Newborn Metabolic Screening Programme

Guidelines for health practitioners

October 2024

Te Kāwanatanga o Aotearoa
New Zealand Government

Health New Zealand
Te Whatu Ora

Guideline

National

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Te Whatu Ora

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He taonga te tamaiti

Every child is a treasure

Contents

Foreword

Health New Zealand |Te Whatu Ora[1] (Health NZ) is responsible for the development, implementation, and management of two newborn screening programmes:

- Newborn Metabolic Screening Programme
- Universal Newborn Hearing Screening and Early Intervention Programme

Health NZ is also responsible for the introduction of quality improvements to Antenatal Screening for Down syndrome and other conditions.

The name of the programme includes the word 'metabolic' however the programme has expanded over the years to screen for a range of disorders; metabolic, endocrine, and others. Most of the screened conditions are inherited (aside from Congenital Hypothyroidism (CH) which can be either inherited or sporadic). For the purposes of this guidance the process will be referred to as newborn blood spot screening and the formal programme referred to as the Newborn Metabolic Screening Programme (NMSP).

While all whānau are advised about screening, participation is optional. Newborn blood spot screening is strongly recommended by Health NZ and occurs with the consent of whānau. Information about newborn blood spot screening should be offered to all whānau to enable them to make an informed decision, and to receive screening results for their pēpi to help inform and plan accordingly.

The purpose of the Newborn Metabolic Screening Programme (NMSP) is to reduce newborn morbidity and mortality through high-quality screening that facilitates early detection and treatment of specific metabolic and other disorders in pre-symptomatic pēpi. This service is offered to all pēpi born in Aotearoa New Zealand.

The success of the programme depends on the diligence and dedication of many health professionals and Lead Maternity Carers (LMCs) in particular. Their input is integral to the continuation of a high-quality screening programme.

These guidelines replace the *Guidelines for practitioners providing services within the Newborn Metabolic Screening Programme in New Zealand* dated 2010.

These guidelines should be read in conjunction with the NMSP policy framework and monitoring indicators, and education resources produced by Health NZ and available on the Health NZ website at Newborn Metabolic Screening Programme – Health New Zealand | Te Whatu Ora.

Contact details for support services and sources of further information about newborn metabolic screening are listed in Appendix One: Contacts and resources.

[1] What was previously known as the National Screening Unit (NSU) has now been incorporated into the Prevention Directorate within the National Public Health Service of Health New Zealand.

Acknowledgements

These Guidelines were developed in 2010 in consultation with the Newborn Metabolic Screening Programme Technical Working Group and were updated in 2023.

Health NZ thanks the Review Panel and the many individuals and groups who contributed feedback to drafts of this document.

Key messages

1. The Newborn Metabolic Screening Programme (NMSP) offers screening to all pēpi (babies) born in Aotearoa New Zealand and contributes significantly to detection of a wide range of metabolic and inherited conditions. Many of the disorders the NMSP screens for can lead to serious illness or mortality for the child within seven to ten days.
2. All whānau should be informed about the NMSP and process during pregnancy.
3. The optimal time for the newborn blood spot collection is 24-48 hrs after birth, or as soon as possible after this (preferably before 72 hrs of age) to prevent irreversible damage and life-threatening illnesses.
4. LMCs are responsible for the newborn blood spot screening process, including giving information and advice, offering screening, ensuring informed consent, documenting the process, taking a suitable sample, and following up results.
5. Correct LMC contact details on the blood spot card is vital because LMCs are the contact point for screening results. LMCs must notify the laboratory of any change to their contact details
6. Blood spot specimens must be suitable for testing and should be sent to the laboratory as soon as they are dry, not batched. Requests for repeat samples must be acted on urgently.
7. Parents/guardians can request that residual blood spots be returned after screening, and should be made aware of storage and possible future use of sample including quality assurance processes and approved research.
8. Clear documentation of the screening process must be kept, including consent, decline, and sample information, results and follow up.
9. The LMC is responsible for checking screening results. If there is no result available by the time pēpi are 7-10 days old the LMC should check that the screening lab has received a specimen.
10. If pēpi receives an abnormal screening result, the LMC (or referring practitioner) is responsible for informing the whānau, and for receiving and following further advice from the laboratory or specialist paediatrician.
11. If there are clinical concerns about a pēpi they must be referred to diagnostic and treatment services without waiting for screening results.
12. No single test checks for everything. No screening test finds all cases of a condition.

List of abbreviations

Abbreviation	Phrase
CAH	Congenital Adrenal Hyperplasia
CF	Cystic Fibrosis
CH	Congenital Hypothyroidism
FAOD	Fatty Acid Oxidation Disorder
GP	General Practitioner
HIPC	Health Information Privacy Code
LMC	Lead Maternity Carer
MCAD	Medium chain Acyl-CoA Dehydrogenase
MSUD	Maple Syrup Urine Disease
NICU	Neonatal Intensive Care Unit
NMSP	Newborn Metabolic Screening Programme
PKU	Phenylketonuria
SCBU	Special Care Baby Unit
SCID	Severe Combined Immune Deficiency
SMA	Spinal Muscular Atrophy

Glossary

Word	Explanation
Congenital	Present from birth
Inherited	Derived genetically from one's parents or ancestors
Sporadic	Occurs for no known reason

Glossary of Māori terms[2]

Te reo Māori	English
Aotearoa	New Zealand
Hapu	subtribe, being pregnant
Hapūtanga	pregnancy
Hui	gathering, meeting, discussion
Iwi	tribe
Kaitiakitanga	guardianship, including stewardship; the processes and practices of looking after the environment
Kārakia	prayer, incantation, blessing
Kaupapa Māori	a philosophical doctrine incorporating the knowledge, skills, and values of Māori
Koha	gift, present, offering, donation
Kōpū	belly, womb, abdomen
Kōrero	conversation, narrative, speech, discourse
Kuia	elderly woman, grandmother, grand aunt
Kupu	word
Mana	inherent authority and dignity
Mana motuhake	an individual's authority to determine their own destiny, self-determination
Mana Wāhine	an approach that privileges Māori women, heritage, and prestige
Mana Whenua	the people of the land who have mana or customary authority; their historical, cultural, and genealogical heritage are attached to the land and sea
Māori	Indigenous New Zealander, Indigenous person of Aotearoa
Mātauranga	knowledge
Mauri	life force
Oriori	birth chants
Papatūānuku	Mother Earth

[2] Moorfield, J.C. (2024) Te Aka Māori Dictionary.

Te reo Māori	English
Purākau	stories
Pēpē	baby/babies (South of Puketutu (Bombay))
Pēpi	baby/babies (Northern regions)
Rangatahi	youth
Taonga	a treasure
Tangata whenua	Indigenous people of the land
Tapu	sacred
Te Tiriti o Waitangi	The Treaty of Waitangi, which is the document upon which the British and Māori agreed to found a nation state and build a government
Te reo Māori	the Māori language
Tikanga	cultural practice, Māori protocols
Tinana	the body, main part of something or someone
Tupuna/Tūpuna	ancestor(s), grandparent(s); has the same meaning as Tipuna/Tīpuna (pl)
Wahine	woman
Wāhine	women
Whakapapa	ancestral lineage
Whānau	family, the smallest social unit of Māori groupings; to be born/give birth. Wāhine hapū are whānau as they move through pregnancy. Also used to refer to parents/guardians. **In regard to informed consent, results, uses of and return of residual blood spots, whānau refers to parents/guardians only.**
Whenua	land, country, earth, ground; placenta

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Editorial Language

In commitment to tāngata whenua and te Tiriti o Waitangi, te reo Māori is prioritised in this document. To maintain narrative flow, the editorial style will refer to wāhine, pēpē/pēpi, and whānau. These terms encompass all priority groups and ethnicities.

Health NZ acknowledges and respects gender diversity within the birthing population of Aotearoa New Zealand, including trans and non-binary people. Culturally safe practice includes health practitioners respecting and engaging with each individual receiving care and adapting their use of language accordingly in practice.

In a medicolegal context it is essential to have clarity about the specific individuals to whom the health practitioner is providing clinical care. In the context of newborn screening this is the pēpi and the whānau. The foundational principle of informed consent requires the health practitioner to protect and support the right of the individual receiving care to exercise rangatiratanga (self-determination) and mana motuhake (bodily autonomy) when navigating health care decision making.

1 Introduction

1.1 Purpose

The purpose of this guideline is to provide health practitioners with clear, concise, and consistent guidance about newborn blood spot screening for metabolic and inherited disorders in Aotearoa New Zealand. They are intended for all practitioners involved in aspects of newborn blood spot screening.

1.2 Target audience

This guideline is written for health practitioners involved in the healthcare of newborn pēpi in Aotearoa New Zealand. Health practitioners should use it to support clinical judgement, knowledge, and expertise, and provide for a timely, consistent, and effective approach to offering and undertaking newborn blood spot screening. Screening participants and their whānau can use this guideline to understand the screening pathway.

The target audience includes, but is not limited to:

- Midwives
- Lead Maternity Carers
- Nurses
- Paediatricians
- Neonatal staff
- Phlebotomists
- General Practitioners
- Laboratory staff
- Obstetricians

1.3 Guiding Principles

- **Informed choice:** The decision to engage in screening is a personal one and the whānau have the right to make an informed choice to accept or decline screening for their pēpi. Informed consent is a foundational principle of the NMSP.
- **Health consumer rights:** The Code of Health and Disability Services Consumers' Rights provides that Aotearoa New Zealand healthcare consumers have a legal right to appropriate information to enable them to give informed consent. During the screening process, the health practitioner is responsible for providing information and education about newborn screening, offering screening appropriately, and receiving and communicating screening results.

- **Cultural safety:** Health services should be tailored to meet the health needs of each individual, recognising the responsibility to support health equity for all, including Māori and Pacific Peoples. Health practitioners should recognise that what works for different populations varies and can familiarise themselves with current health strategies and health plans developed by Health NZ and the Ministry of Health. See also Te Tatau o te Whare Kahu | Midwifery Council Statement on Cultural Competence for Midwives.
- **Equity of access:** Health practitioners should be aware that barriers to accessing aspects of postnatal care and screening may include lack of knowledge, mistrust of health services, lack of access to postnatal care services, and diverse cultural views of health.
- **Rights of disabled persons:** Health practitioners should offer additional support to whānau who have difficulty understanding information because of language difficulties, hearing impairment, or intellectual disability. Further information can be found on the Whaikaha (Ministry for Disabled People NZ) website.
- **Protecting privacy:** The Health Information Privacy Code 2020 (HIPC) sets specific rules for agencies in the health sector to ensure the protection of individual privacy. It addresses health information collected, used, held, and disclosed by health agencies. The HIPC requires agencies to be clear about the purpose for which they collect information, and open about those purposes to the health consumers from whom it is collected. Health information must be held securely to protect it against misuse, loss, or unauthorised disclosure. Health consumers (or, for pēpi and rangatahi, their parents) can access their health information (with some minor exceptions) and seek its correction when it is wrong. Health information should only be used or disclosed for the purposes for which it was collected, unless one of the exceptions in the HIPC applies.
- **Professional accountability:** Health practitioners providing lead maternity care have an obligation under the Primary Maternity Services Notice 2021, issued pursuant to Section 94 of the Pae Ora (Healthy Futures) Act 2022, to advise whānau of screening services available that are endorsed by Health NZ, including the Newborn Metabolic Screening Programme.
- **Legislation:** Health practitioners are responsible for documentation of screening discussions and choices in the clinical notes, ensuring compliance with the:
    - Privacy Act 2020 and Health Information Privacy Code 2020
    - Code of Health and Disability Services Consumers' Rights 1996
    - Health Act 1956
- Health Practitioners Competence Assurance Act 2003

- Public Records Act 2005
- Pae Ora (Healthy Futures) Act 2022

Other screening resources are available for health practitioners at Health New Zealand | Te Whatu Ora.

1.4 Te Tiriti o Waitangi

Our Te Tiriti o Waitangi obligations are fundamental to screening initiatives in Aotearoa New Zealand. This requires a strong focus on the principles found in Te Tiriti o Waitangi and must align with the transformations called for in the Wai 2575 Hauora report of the Waitangi Tribunal. As identified in the Hauora report, the Waitangi Tribunal proposed the framework of Te Tiriti o Waitangi principles be adopted for the primary health care system, inclusive of tino rangatiratanga, equity, active protection, options, and partnership. These principles can also be adopted for population-based screening programmes.

The Waitangi Tribunal concluded that persistent health inequities that Māori experience were the consequence of the failure to apply the principles of Te Tiriti at structural, organisational and health practitioner levels of the health and disability sector. Giving effect to Te Tiriti requires health practitioners to know the principles of Te Tiriti and to capably apply these in partnership with Māori in their day-to-day maternity clinical practice. Applying the principles to maternity service delivery is vital to enabling Māori to express their mana[3] and to receive high-quality, culturally safe care, and achieve equitable health outcomes.

How these principles apply to health services is supported by *Ngā paerewa* and, in particular, Pae ora healthy futures.

For the health and disability sector, the principles of Te Tiriti are as follows:

- **Tino rangatiratanga**: Health practitioners support the right of Māori to receive effective health care, conceptualising the decisions of the whānau as a continuation of a much older, Māori collective-endorsed practice of self-determining one's own health and wellbeing and that of the whānau.
- **Equity**: Health practitioners can contribute to equitable health outcomes for Māori by ensuring that, at a minimum, their health outcomes match those of other New Zealanders. Equitable outcomes will be achieved when health practitioners work in ways that give effect to the principles of Te Tiriti and *Ngā paerewa*. The principle of equity requires the Crown to commit to achieving equitable health outcomes for Māori.

[3] See Ministry of Health's Te Tiriti o Waitangi Framework for the Ministry's four goals, each expressed in terms of mana. URL: https://www.health.govt.nz/system/files/documents/pages/whakamaua-tiriti-o-waitangi-framework-a3-aug20.pdf (accessed 2 February 2022).

- **Active protection**: Health practitioners should share evidence-based information about maternity and newborn outcomes so that Māori can make decisions and prepare themselves to uphold their tikanga (for example, karakia, rongoā, support people). Health practitioners must actively support Māori to make decisions that are best for them.
- **Options**: Health practitioners must ensure Māori have access to health care that enables them to uphold their tikanga regardless of where care is being provided. Processes must complement a Māori person's mana, support their tikanga, and be culturally safe as defined by Māori.
- **Partnership**: Health practitioners work in partnership with Māori, including their whānau as defined by them. A partnered approach to the process and decision-making ensures Māori can enact their rangatiratanga while exercising mana motuhake (authority over their bodies and reproductive health).

1.5 Equity

In Aotearoa New Zealand, people have differences in health outcomes that are not only avoidable but unfair and unjust.[4] Differences in the structural determinants of health and wellbeing (for example, disadvantages in income, employment, education, and housing, as well as multiple forms of discrimination) negatively impact people's health but people have little control over these. Health inequities are not about people; instead they are the result of avoidable structural determinants in our communities.[5] When health practitioners understand the structures that create inequitable maternity outcomes, they can use different approaches and resources to achieve equitable health outcomes.

Achieving equitable health outcomes for all happens when health service providers and health practitioners:

- understand the structures that create disadvantage for those groups.
- are supported to work in ways that give effect to the principles of Te Tiriti, as well as meeting professional competencies and *Ngā paerewa*.

Lastly, health practitioners should be aware that many people in Aotearoa New Zealand conceptualise anatomy, pregnancy, gender, sexuality, reproduction, contraception, and birth in diverse ways according to their worldviews. Therefore, health practitioners should use proven health literacy practices[6] to communicate effectively with everyone using their services (for sector guidance, see *Ngā paerewa* Standard 1.4 E whakautetia ana ahau | I am treated with respect and criteria 1.4.2).

[4] Ministry of Health. 2019. *Achieving Equity*. URL: https://www.health.govt.nz/about-ministry/what-we-do/work-programme-2019-20/achieving-equity (accessed 2 February 2022).
[5] Toi Te Ora Public Health. 2021. *Determinants of Health and Health Equity*. URL: https://toiteora.govt.nz/public/determinants-of-health-and-health-equity/ (accessed 2 February 2022).
[6] Ministry of Health. 2015. A framework for health literacy. URL: https://www.health.govt.nz/publication/framework-health-literacy (accessed 2 February 2022).

2 Background information

2.1 Overview

The NMSP is one of the most successful screening programmes in Aotearoa New Zealand. Almost all pēpi born in Aotearoa New Zealand are screened, and as a result approximately 60 pēpi are identified with and treated for a screened disorder each year. When serious disorders are diagnosed in early infancy, treatment can commence immediately, preventing irreversible damage and life-threatening illnesses.

Newborn blood spot screening involves collecting a blood specimen from pēpi (the 'heel prick test') onto a blood spot card (sometimes called a 'Guthrie card'). The blood samples are tested for metabolic and other inherited disorders in a laboratory.

Health NZ has responsibility for the funding, monitoring, and strategic direction of the NMSP. The screening laboratory and follow-up programmes are run by LabPLUS, Te Toka Tumai Auckland, Health NZ.

Potential benefits and harms of screening

For those being screened:

- That early detection of the disorders screened for can enable prompt treatment.
- That the disorders screened for cannot be easily detected without blood tests.
- That the disorders screened for can be life-threatening.
- That screening is not diagnostic, and there is a possibility of an abnormal screening result in unaffected pēpi and a normal screening result in affected pēpi.
- That whānau may receive insufficient and/or inappropriate information to allow them to make an informed choice about screening.

For pēpi with abnormal results:

- Potential morbidity associated with delayed diagnosis if samples are unsuitable, taken late or delayed in transit to the laboratory.
- Potential inequities associated with lack of access to specialist metabolic or paediatric services.

At a societal level:

- Timely screening can reduce the cost of managing the health of individuals with morbidity associated with metabolic and inborn disorders (offset by the comparatively low cost of the screening programme).
- The possibility that practitioners may ignore clinical symptoms caused by a disorder if a screening result is normal.

- There may be parental anxiety associated with waiting for results.
- There are potential future benefits from the use of residual blood spots, which are currently stored on a long-term basis.

2.2 A history of newborn blood spot screening in Aotearoa New Zealand

Aotearoa New Zealand was one of the first countries in the world to implement a national newborn metabolic screening programme, commencing screening in 1969. Screening was initially only for phenylketonuria (PKU). As technology improved, tests for maple syrup urine disease (MSUD), galactosaemia, histidinaemia and homocystinuria were introduced. In 1978 a test for congenital hypothyroidism (CH) was added, and in 1979 scientists in Aotearoa New Zealand developed immunoreactive trypsin as a marker for cystic fibrosis (CF), and CF screening was run as a research pilot project before being added to the programme in 1986. Congenital adrenal hyperplasia (CAH) and biotinidase deficiency were also added in the 1980s. Screening for homocystinuria and histidinaemia was stopped in the early 1980s as a result of monitoring the efficacy of the programme.

In 2006, the gifting of a tandem mass spectrometer by the Starship Foundation allowed the NMSP to increase screening from seven to over 20 disorders, with the addition of 9 fatty acid oxidation disorders (FAODs) and 12 more amino acid disorders. A video celebrating the 50th anniversary of the Newborn Blood Screening Programme and short videos of affected whānau talking about what newborn screening has meant for their child is available to watch at Tell us your story - Celebrating 50 years of newborn metabolic screening (info.health.nz). The next significant additions to the programme were the introduction of screening for Severe Combined Immunodeficiency (SCID), an inborn error of immunity, in 2017 and Spinal Muscular Atrophy (SMA) on 12 February 2025.

2.3 Disorders in the Newborn Metabolic Screening Programme

The disorders currently screened for by the NMSP are:

Metabolic:

- Amino acid disorders (including PKU and MSUD)
- Fatty acid oxidation disorders
- Galactosaemia
- Biotinidase deficiency

Other:

- Congenital hypothyroidism
- Cystic fibrosis

- Congenital adrenal hyperplasia
- Severe Combined Immune Deficiency (SCID)
- Spinal Muscular Atrophy (SMA)

For more information on these disorders see Appendix Two: The Screened Conditions.

3 Te Ao Māori

Within a Māori worldview screening and the sampling of Māori DNA (and its association to whakapapa and te ao Māori) have important cultural values and hold a sacred significance for Māori. Wāhine and their ability to give birth have special significance in te ao Māori. This ability to continue whakapapa is celebrated, making childbirth one of the most important traditions in society. In Māoridom the practice of oriori while the child is in the womb is seen as an important aspect of connecting to whakapapa and acknowledging the past, present, and future aspirations for the child growing within the womb. Traditional Māori birthing practices focus on the importance of conveying ancestral journeys, stories, and achievements of ancestors through karakia and oriori. This practice gives the unborn child something to aspire to, and encourages a lifelong pursuit of learning, prosperity, and aroha for pēpi and whānau. This grounds the child through whakapapa to the land, and to papatūānuku. The Tuku Iho app is a useful resource for discovering oriori and kaupapa Māori antenatal resources.

Maternal figures are prominent in Māori cosmology and kōrero tuku iho (narratives passed down by Māori through oral tradition). These pūrākau (stories) form the basis of many tikanga pertaining to wāhine Māori and their birthing rites.

Health practitioners in Aotearoa New Zealand should ensure they are aware of the cultural importance of blood for Māori and its significant association to whakapapa. Blood is considered tapu (sacred) in te ao Māori and the storage or return of samples collected for screening must be fully explained with options provided for returning the sample to the whānau.

A Māori perspective on ‘disability’ may focus on the unique strengths of the person affected rather than a deficit mindset. Health practitioners should ensure they are aware of potential cultural differences for any whānau and choose their language carefully when counselling whānau.

For more information see:

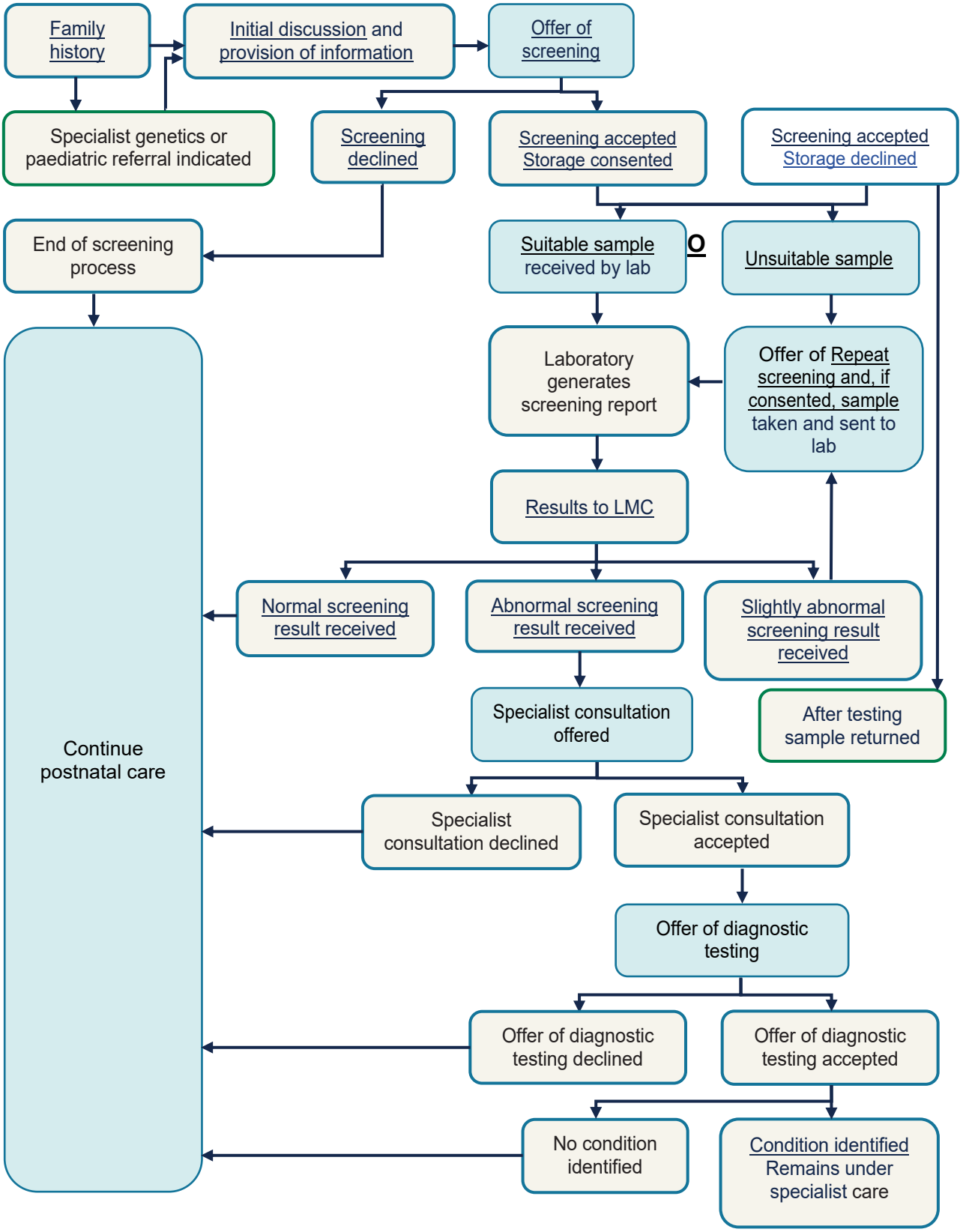
Hakui website

Turanga Kaupapa

Tuku Iho app

Whānau hauā: Reframing disability from an Indigenous perspective

4 The screening pathway



4.1 The screening process

The screening process starts with the provision of information to whānau during pregnancy and includes an initial discussion about screening, an offer to screen, collection of a blood sample, laboratory testing, and follow up where required. The process ends when pēpi receives a normal screening result or is referred for diagnostic testing, or if the whānau withdraws from the process.

When laboratory testing is complete, the residual blood spots are either held in secure storage indefinitely or returned to whānau (see Storage, return, and future uses of blood spots). Stored blood spots may be used in the future for the purposes listed in Uses of residual blood spots after screening.

Whānau (or the individual) can request to have the blood spots returned from storage.

5 Screening discussion

5.1 Family History

If any of the disorders screened for are present in the whānau, pēpi may have an increased chance of inheriting the disorder. It may be appropriate for pēpi to undergo prenatal or cord blood diagnostic testing as well as being screened. LMCs should refer whānau with a family history of disorders to a paediatrician or geneticist for further advice during pregnancy.

For example, if there is a family history of CF, consultation with a CF paediatrician or genetic services as soon as possible (ideally when planning the pregnancy) is recommended. If pēpi is born without prior testing and has a normal screening result a consultation with a CF paediatrician is still recommended as pēpi severely affected by CF can have normal screening results.

5.2 Screening discussion

Health practitioners should offer additional support to whānau who have difficulty understanding information because of language difficulties, hearing impairment or intellectual disability. Linguistically diverse whānau may require access to interpreter services to ensure they understand the information provided to them about newborn blood spot screening. Using friends or whānau members as interpreters is not recommended practice. Interpreter services are funded by Health NZ using Connecting Now. To access Connecting Now's interpreting service the phone number is 0800 854 737 (PIN 14059) or you can email support@connectingnow.com.au.

The discussion must include the following information:

About the NMSP

- The purpose of screening (ie, how screening can help pēpi who have metabolic or other inherited disorders).
- That the screening tests are for defined, serious disorders.
- That screening does not cover every metabolic or inherited disorder.
- What screening involves: sample taking and testing, data and information collection and monitoring, reporting and follow-up of results, quality assurance processes, referrals for diagnostic testing and treatment in the case of positive results, and storage and possible future uses.

Resources

- Availability of consumer resources including 'Your newborn baby's blood test'. **This should be given at the time of the discussion**.
- Information and resources to assist with screening discussions are listed in Appendix One: Contacts and resources.

Consent

- That screening is voluntary.
- That whānau may decline screening on behalf of pēpi.
- That if a screening result is abnormal, the screening laboratory will either request a further blood spot sample, or a specialist paediatrician will contact the whānau for a consultation and arrange for pēpi to be assessed and have diagnostic testing.
- That consent is required for screening and for storage and for possible future uses of residual blood spots.
- The right of whānau to withdraw consent if they change their minds at any stage of the screening process until the sample has been sent to the laboratory.
- The right of whānau to accept screening after initially declining.

Storage, return, and future uses of residual blood spots

- That screening can be undertaken without the sample being retained by the laboratory (ie, residual blood spots can be returned to the whānau).
- How long residual blood spots are stored for.
- Possible uses of residual blood spots, including improving programme quality.
- Security of and access to stored blood spots.
- The right of the whānau, or the individual, to request return of the residual blood spots at any time.

Sample taking

- How the blood sample is taken.
- How whānau can prepare pēpi for the test and comfort them during sample taking.
- That **a repeat sample is needed for about one in every 100 pēpi.**

Results

- That all results are notified to LMCs.
- That unsuitable samples will be notified to the LMC.
- That abnormal results that indicate a probable disorder will be phoned to the LMC by a senior/clinical scientist. Some abnormal results need urgent clinical intervention.

Data and information collection and monitoring

- The information and data collected about māmā and pēpi and what this information and data may be used for.
- That the NMSP is monitored at a national level.

6 Documentation

Clear documentation of the screening process must be kept, including consents, declines, and sample information.

Written consent for newborn blood spot screening is not required by the Code of Health and Disability Services Consumers' Rights. However, details must be documented in the clinical records. This is the responsibility of LMC's, or whoever the LMC delegates to collect the sample, for example hospital phlebotomy. If there is no LMC assigned, the postnatal care provider is responsible.

Each stage of the process should be documented in the clinical records, including:

- the content of discussions about the NMSP, any further information requested, issues raised, and/or resources provided
- the use of interpreters or similar services
- consent or decline for screening
- consent or decline for storage and possible future uses of residual blood spots
- the date and time the sample was taken
- details of repeat samples, follow-up, and referrals in the case of abnormal results

Handover notes to Well Child/Tamariki Ora providers, general practitioners (GPs) and other health care providers must include:

- documentation about the screening process
- consent or decline for screening
- details about the blood sample (including the date taken and date sent)
- results of screening
- any follow-up from screening results

7 Informed consent

7.1 Informed consent requirements

Ensuring that whānau give informed consent for newborn blood spot screening is a legal requirement.

Informed consent is a process that must be integrated throughout the screening pathway. Ensuring informed consent includes:

- provision of information about screening during antenatal and postnatal care
- discussions about screening throughout antenatal and postnatal care
- offering screening
- documenting consent or decline to screening
- documenting consent or decline to storage and possible future use of blood spot samples

8 Offer of screening

8.1 Responsibility for screening offer

LMCs are responsible for the newborn blood spot screening process, including giving information and advice, offering screening, ensuring informed consent, documenting the process, taking a suitable sample, and following up results.

If there is no LMC assigned for maternity care, the primary maternity health provider or the secondary/tertiary service is responsible for the screening process.

Whānau must decide:

- whether they agree to their pēpi being screened for metabolic and inherited disorders
- if they agree to screening, whether they agree to the residual blood spots being stored for possible future uses or want the residual blood spots returned after screening

Care providers should make it clear to whānau that they have two separate decisions to make and facilitate informed choices for each.

Whilst all pēpi born in Aotearoa New Zealand are offered blood spot screening through the NMSP, it is important that health practitioners inform whānau who are not eligible for funded health care that there may be costs involved in diagnostic testing, treatment, and ongoing care of any affected pēpi.

8.2 When screening is declined

Health NZ strongly recommends that all pēpi are screened.

However, whānau have the right to accept or decline the screening offer. If screening is declined, LMCs should document the decision in the clinical records and handover notes to General Practice, Well Child/Tamariki Ora providers and other health care providers. LMCs should remind the whānau that they can have screening done at any time if they change their mind.

If whānau agree, the card should be filled in with the demographic information for pēpi, LMC details, and a note that screening was declined, and sent to the laboratory to monitor participation in the NMSP.

9 Sample taking

9.1 Responsibility for taking samples

LMCs are responsible for taking a suitable sample within the recommended timeframe and ensuring timely transport to the laboratory. The responsibility for sample collection may be delegated, for example to a hospital service or post-natal facility, who then assumes responsibility for transport to the laboratory.

If there is no LMC assigned for maternity care, the postnatal care provider is responsible for ensuring a suitable sample is taken within the recommended timeframe and ensuring timely transport to the laboratory.

If care has been transferred to a secondary/tertiary service, then that service is responsible for ensuring a suitable sample is taken within the recommended timeframe and ensuring timely transport to the laboratory.

9.2 Sample takers' responsibilities

The sampling practitioner must:

- check that the whānau have given consent for the screening test
- collect an adequate sample
- document accordingly

If whānau have not received information and advice about the NMSP (for example due to late presentation to maternity services, or transfer of care), or do not understand the purpose of newborn blood spot screening, the sampling practitioner should not take the sample, and should inform the LMC or postnatal care provider as soon as possible that the sample has not been taken, and the reason.

9.3 Timing of taking samples

The optimal time for sample collection is 24 - 48 hours after birth (or as soon as possible after this).

Taking samples when pēpi is 24 hours old or as soon as possible after this allows early diagnosis and treatment if a disorder is present.

Severe forms of some of the disorders screened for can be fatal within seven to ten days of age but pēpi may not show any signs or symptoms until irreversible damage has occurred. For example, pēpi affected by serious metabolic disorders can be born without any signs or symptoms because the placenta eliminated abnormal and harmful biochemicals produced by the baby's system while pēpi was in utero.

For effective newborn blood spot screening, pēpi must have been independent from the placental circulation long enough for many of the indicator markers to show an abnormality. In pēpi with fatty acid oxidation disorders (FAODs), the catabolic state all pēpi experience in the first days after birth changes the indicator fatty acid markers. These markers normalise when pēpi starts processing larger volumes of milk. In other disorders (for example, amino acid breakdown disorders) the markers will continue to be abnormal, and some will rise to toxic levels.

The sample must be taken late enough for the disorders to be detectable, and early enough for the fatty acid oxidation indicators still to be high. **The earliest optimal window of opportunity is now understood to be between 24 and 48 hours.**

It is possible for samples to be taken later if whānau initially decline screening but then change their minds, even though the optimum time for sample-taking has passed.

Milk feeding, stool colour, and antibiotic use do not affect results and must not delay sample-taking.

9.4 Blood spot cards

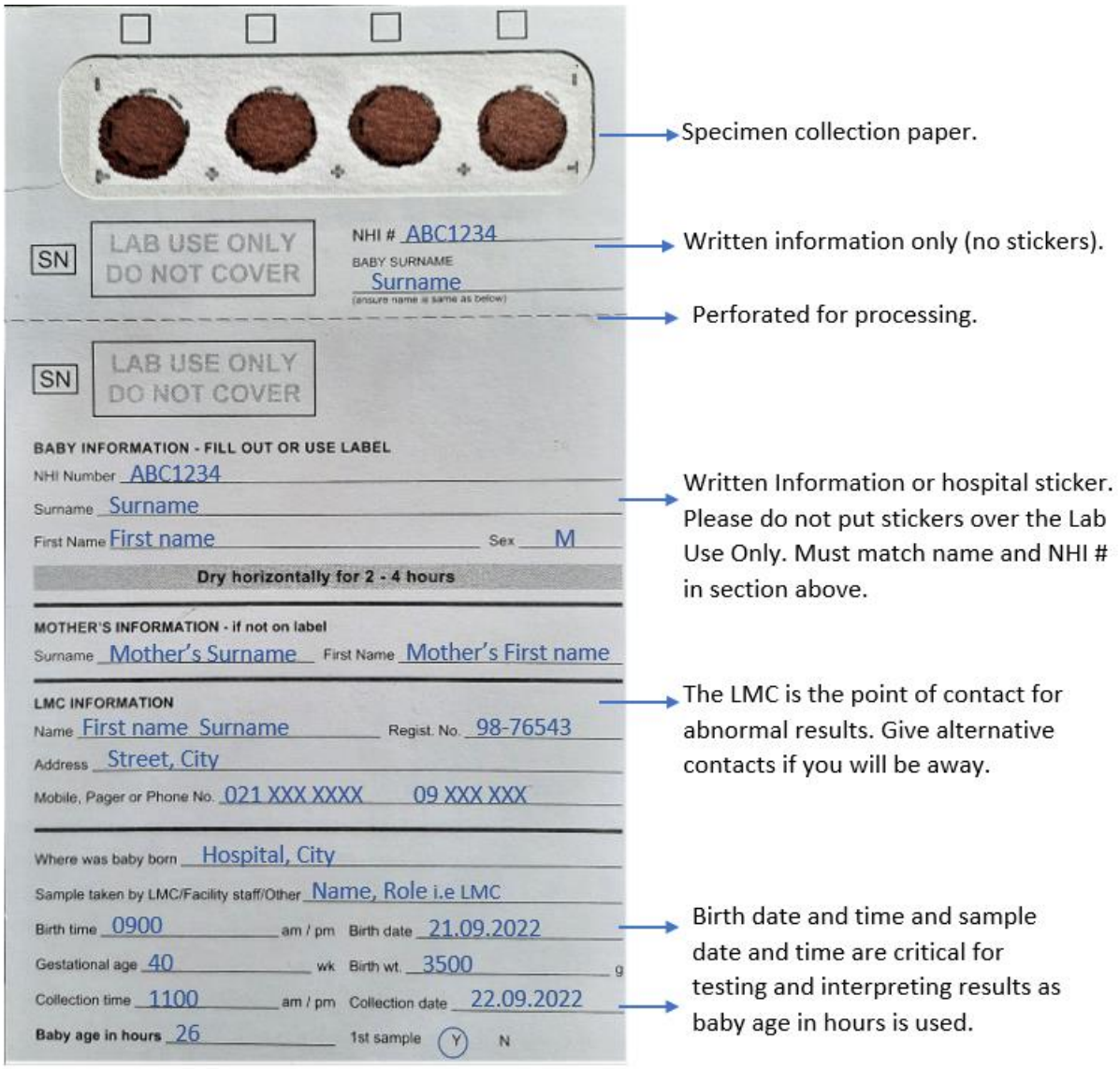
Blood spot cards have two components: a smaller portion with specimen collection paper for the blood sample, and a larger portion for demographic and other information. The two components are separated by a perforation. When the card is received by the laboratory, a unique identification number is placed on both sections of the card and the laboratory separates the blood spot portion from the rest of the card for testing.

Ensuring LMC contact details are on the card is vital because LMCs are the contact point for screening results, and significantly abnormal results must be acted on urgently. LMCs must notify the laboratory of any change to their contact details.

It is essential that the laboratory can identify the pēpi that the sample belongs to. The name of pēpi needs to be written and the same on both the demographic and sample areas of the card. If this is not done (for example because different maternal and paternal surnames are used in different areas), the laboratory will contact the LMC to confirm the identity of pēpi.

- **If the LMC will not be available to be contacted by the laboratory regarding potential abnormal results, the card should be filled in with the contact details of the backup LMC, responsible facility or another available practitioner.**
- Fill in all fields on the card
- Do not use Sellotape or staples on the card
- Courier to lab as soon as it is dry (1 ½ -2 hrs)

Figure 1: Blood spot card



To order, lancets, blood spot cards and pre-addressed courier envelopes for newborn metabolic screening, please email newbornscreeningresources@adhb.govt.nz or call 09 307 4949 extn 23806.

9.5 Procedure for taking samples

An interactive e-learning module has been developed to support best practice in newborn bloodspot sample collection. This is available to complete (free of charge) at Learn Online.

Midwives can attribute completion of this to ongoing education requirements for recertification with the Midwifery Council of New Zealand.

Samples must be suitable for testing.

Figure 2: Correct sample-taking procedure

Note that a separate protocol applies for pēpi born **preterm or a with a birthweight under 1500g.**

1. **Obtain informed consent from whānau.**
   Obtain consent for:
   - screening
   - storage and possible future uses of residual blood spots (see Storage, return, and future uses of residual blood spots)

2. **Fill in all the fields on the card.**
   Fill in the blood spot card prior to and at the time of sample taking. Complete all fields and ensure that a contact telephone number for the LMC (or back up if LMC on leave) is provided in case the laboratory requires an urgent contact for positive results.

3. **Gather equipment.**
   Equipment needed for the procedure:
   - gloves
   - cleansing material (if required)
   - approved lancet (see step 9)
   - blood spot card with written information fields completed

4. **Ensure pēpi is warm and comfortable.**
   Analgesia in the form of breastfeeding, bottle feeding, or skin-to-skin contact is beneficial. Encourage pēpi to breastfeed or be cuddled during the procedure. Sucrose may also be used as pain relief with parental consent[7].

5. **If there is any need to warm the foot, do so with care.**
   A warm heel is needed for good perfusion. Warm with booties and blankets if necessary. Collectors should be aware of the risk of causing serious burn injury through ad hoc pre-warming, for example gloves or nappies filled with hot water.

[7] Royal Australasian College of Physicians: *Pediatrics and Child Health Division. 2005. Guideline Statement: Management of procedure related pain in neonates.* Sydney: Royal Australasian College of Physicians

6. **Use universal infection control precautions, including hand washing and gloves, during sample-taking.**
   Hand washing and the use of gloves during sample-taking provides protection for pēpi from infection and protects the sampling practitioner from blood contamination.
7. **Make sure the foot is clean and dry.**
   If necessary, clean the foot with warm water or an alcohol swab, and allow it to dry before taking the blood sample.
8. **Allow the foot to hang down to aid blood flow.**
9. **Use an NMSP approved lancet.**
   Use an automated lancet with a depth of incision less than or equal to 2.4 mm. Special lancets are available for premature/small pēpi.
   There are a number of approved lancets available in Aotearoa New Zealand and practitioners can access these free of charge via Newborn metabolic screening resources – Health New Zealand | Te Whatu Ora. These are specifically designed for newborn blood sample-taking and should always be used for this purpose. A list of currently approved lancets can be found via the heel prick guide.

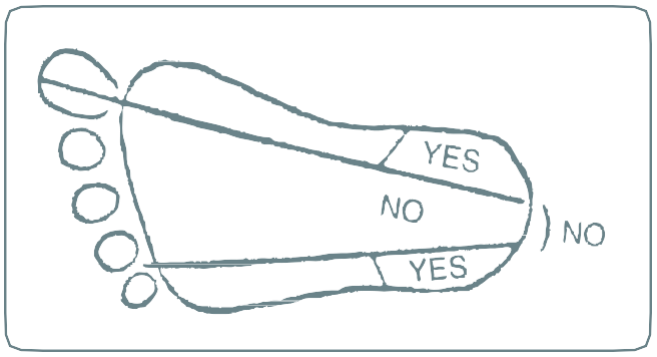
Figure 3: Quikheel™ lancet and Tenderfoot® lancet

Various studies have examined the effectiveness of different lancet types (‘stabbers’ versus ’slicers’). As a result, Health NZ recommends the slicing device, which generally decreases the need for more than one heel prick per test and reduces crying and pain for pēpi.

Place the lancet firmly against the heel in the area shown in the diagram, then activate.

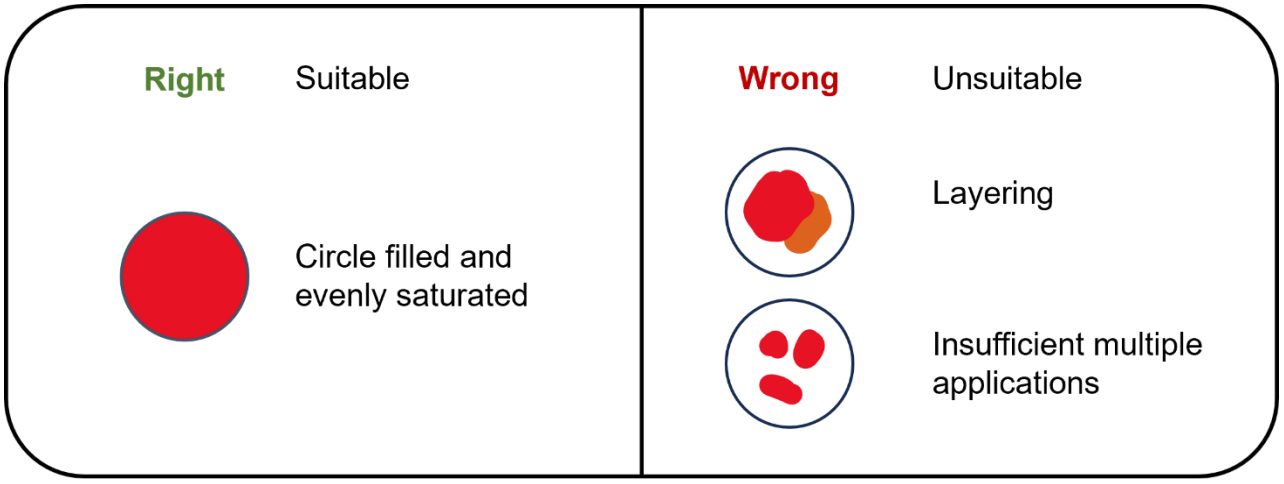
To avoid damage to the calcaneus and risk of osteomyelitis, a sample site should be selected on the most medial or lateral portions of the plantar surface of the heel, and not on the posterior curvature of the heel.

Figure 4: Recommended sample sites



10. **Completely fill each of the four circles on the blood spot card, filling all spots from the same side of the card.**
    - Wait up to 15 seconds for blood to flow.
    - Wipe off the first drop.
    - Allow blood to fill each spot by natural flow.
    - Fill from one side of the card and allow blood to seep through.
    - Ensure each spot is filled completely before moving onto the next.
    - Do not layer spots.
    - Do not touch the collection paper on the blood spot card. If anything comes into contact with the collection paper (for example, dirt, powder, lotions, or creams), it will be contaminated.

Figure 5: Suitable and unsuitable samples



A good sample consists of:

- four well-filled spots (to the dotted line)
- spots filled from one side of the specimen collection paper
- one spot filled at a time
- fully completed blood spot card information
- spots that are dried before posting and not left in the sun, overheated, or posted while wet
- specimen collection paper that has not been contaminated

11. **If a second puncture is required, select a new site.**
    Perform the second puncture on a different part of the same foot, or on the other foot.

12. **Apply gentle pressure with a clean swab to the wound to stop bleeding.**
    Gentle pressure with a swab is usually sufficient to stem the flow of blood.
    If a plaster is required for haemostasis or infection control, use those with easy to remove adhesive (for example micropore).

13. **Dry the card horizontally for 1½–2 hours.**
    Cards must be dried horizontally, out of direct sunlight, and not in a hot car. Spots are dry when they are no longer red.
    When the blood is dry, wrap the cover over to protect the spots.

14. **Send the sample to the laboratory.**
    See Sending samples to laboratory.

15. **Unsuitable sample**
    Sample takers should ensure that samples are taken, dried and sent in accordance with these Guidelines. If samples are unsuitable (see Unsuitable samples), a repeat sample will be needed, and screening will be delayed. If pēpi has one of the disorders screened for, this delay could compromise their access to timely diagnosis and treatment.

9.6 Sending samples to the laboratory

Samples must be sent to the laboratory by courier as soon as they are dry to ensure testing can be completed early enough to give clinical benefit if a disorder is identified. Severe forms of some of the disorders can be fatal within seven to ten days. Blood samples can be compromised if they take too long to reach the laboratory.

- Samples must never be batched over several days for sending, even if they are being sent from a hospital.
- When sending cards by courier, ensure the courier envelope containing the sample card is dropped at a **daily** collection site or call the courier service directly to request

pick up (0800 COURIER (0800 268 743) or the courier service currently used by your facility). Visit Find NZ Post | NZ Post to find your nearest NZ Post drop off point.

- **Do not place courier envelopes in standalone post boxes as these may be infrequently cleared.**
- The sample-taking and sending must be recorded in the clinical notes. It is useful to record the courier tracking number, for example by downloading the NZ Post App and scanning or taking a photo. This can be used to identify the whereabouts of samples where transit is delayed.

Posting of biological materials requires triple containment. The first containment is the collection paper fibres, the second is the card wrap and the third is the mailing envelope.

To order pre-addressed courier envelopes, lancets, and blood spot cards for newborn metabolic screening, please email newbornscreeningresources@adhb.govt.nz or call 09 307 4949 extn 23806.

9.7 Use of capillary tubes and arterial lines

Health NZ does not recommend capillary tubes for newborn blood sample collection. International studies indicate that they may cause haemolysis or microtears in the specimen collection paper.

Capillary tubes with anticoagulant must not be used.

Blood samples may be taken from venous and flushed arterial lines if these are in place.

9.8 Low birth weight and sick pēpi, and effect of blood transfusions

Low birth weight and sick pēpi often receive false positive results due to their immaturity and/or illness. Blood transfusions can also affect test results. Protocols have been

developed to ensure effective screening for these pēpi (see Appendix Four: preterm metabolic bloodspot screening protocol and Appendix Five: Newborn metabolic screening protocol for babies who have received blood transfusions).

Requests for repeat samples must be acted on urgently.

The laboratory may request a repeat sample if the first sample is unsuitable, or testing shows slightly abnormal results. A repeat sample is required for about one in every 100 pēpi.

For more information about unsuitable samples and abnormal results, see Screening results.

The laboratory will advise the LMC (or practitioner identified on the sample card) why the sample was unsuitable or what the abnormal results indicated.

If the sample is unsuitable, the laboratory will either phone the LMC or send an SMS text message requesting a further blood spot sample, advising that the initial sample was unsuitable and could not be fully tested, and the reasons for this.

If the result indicates that a disorder is probable, a specialist paediatrician associated with the screening programme or senior scientist will phone the LMC to discuss the clinical situation for pēpi and advise of next steps.

If the result is unclear or indicates a possible mild form of one of the disorders, a laboratory senior scientist will telephone the LMC requesting a further blood spot sample. Where possible, the LMC will also be texted a link to an information sheet about the possible disorder and other reasons for the abnormal result.

Further information about specific disorders is in Appendix Two: Disorders in the Newborn Metabolic Screening Programme.

LMCs must provide whānau with sufficient information about the need for a repeat sample and follow the repeat sample procedure below, having regard to the level of urgency advised by the laboratory.

9.9 Unsuitable samples

If a sample is unsuitable for testing a correct screening result will not be able to be provided. Pēpi will not have been fully screened for metabolic and other inherited disorders. Samples may be unsuitable due to:

- insufficient blood
- blood being layered onto a spot (too much blood)
- blood spots being squeezed to push the blood through
- contamination from dirt, powder, lotions, creams, skin oils or anything else coming into contact with the specimen collection paper
- delays in the card arriving at the laboratory

- the card wrap being folded over the blood spots before the blood is dry
- cards being posted before the blood is dry (wet samples may go mouldy)
- exposure to heat (for example, after being left in a hot car)

If a sample is unsuitable, the laboratory will contact the LMC via a phone call or text message to request a repeat sample. A repeat sample must be provided so that pēpi can be screened.

Note that the laboratory still tests unsuitable screening samples as some markers may be informative and this can allow for earlier notification of time critical results. If this occurs, the LMC will be notified as for other disorder probable results but will be advised that a further sample is still needed to complete routine screening.

The LMC is responsible for taking the repeat sample and must follow the procedure in Repeat sample procedure.

9.10 Repeat sample procedure

1. **Contact the whānau**
   - Contact the whānau and explain why a repeat sample is necessary.
   - If sample results were abnormal, advise the whānau of the results.
   - If pēpi is unwell, refer to a GP, paediatrician, urgent afterhours services or the emergency department.

   If the LMC is unable to contact the whānau:
   - the laboratory must be notified.
   - the GP or Well Child/Tamariki Ora provider should also be notified.
2. **Ensure informed consent**
   Confirm consent before taking repeat samples. Consent must be documented separately for:
   - screening.
   - storage and possible future uses of residual blood spots.

   If the repeat sample is declined, notify the laboratory by emailing NMSP@adhb.govt.nz
3. **Take the repeat sample in accordance with guidelines for the first sample.**
   (see Procedure for taking samples).
4. **Send the repeat sample to the laboratory as soon as the blood is dry.**
   The monitoring standard is that further blood spot samples are received in the laboratory (or decline notified) within 10 days of the laboratory request for a repeat sample.
5. **Notify whānau of the repeat sample results when they are received.**

6. If no repeat sample is received by the laboratory, the lab will:

- Follow up requests for repeat samples by phone/text.
- Send a reminder after one week.
- Send a second reminder after two weeks.
- Re-send a formal report to LMC.
- Send a final reminder after four weeks.

Reminders will ask for progress on collection of a sample, and this is where LMCs can advise of difficulty contacting the whānau.

10 Results

10.1 Results

There are three possible screening results:

- condition unlikely (normal test)
- unsuitable sample (with reason)
- condition possible (borderline) or probable (abnormal test)

LMCs must reconcile laboratory reports with samples they have sent for testing and clarify any discrepancies with the laboratory. This includes screening samples collected on behalf of the LMC or whilst pēpi was still in hospital or a birthing centre.

LMCs will receive results for every sample collected and tested. The laboratory keeps records of requestor contact details and their preferred method for receiving reports. Newborn screening reports are increasingly sent electronically rather than mailed as hard copies and are no longer faxed. LMCs should contact 0800 LABPLUS to notify of a change of contact details, including to update their mobile phone number or change their reporting preference. **The lab will use text messaging** to communicate at times (unless LMCs have opted out of this contact method), however formal reports will always be sent to the LMC via usual method. Text messages are computer generated so can be replied to, but the number cannot be called back. The message will contain a number to ring to speak to a person.

If there is no screening result available by the time pēpi is 7-10 days old, the LMC should call 0800 LABPLUS to check that the sample has been received by the laboratory. If the courier tracking number was retained the LMC can also follow up with the courier company. Prior to LMC discharge, the LMC should check that they have received a screening result.

If there are clinical concerns about pēpi, they must be referred to diagnostic and treatment services without waiting for screening results.

If an LMC has clinical concerns about pēpi, they must refer them to a GP, paediatrician, acute after-hours services or the emergency department regardless of the screening result or whether a result has been received.

Screening results are confidential. Access to the results is restricted to those involved in the NMSP and those caring for pēpi.

10.2 Normal results

A normal screening result means that marker levels are within the normal range and pēpi has a very low chance of having one of the disorders screened for.

10.3 Abnormal results

All abnormal results that are suggestive of a disorder will be communicated as a phone call to the LMC, using the contact number recorded on the screening card.

If an abnormal screening result is received, the LMC must take further action, according to the advice of the laboratory or specialist paediatrician associated with the screening programme.

The LMC is responsible for communicating abnormal screening results to whānau. Abnormal screening results are ideally communicated to whānau in-person, using written information from the laboratory or a specialist paediatrician.

There are different categories of abnormal screening test results.

If the abnormal result suggests pēpi very likely has a disorder (which may be life-threatening) the call will be made by a specialist paediatrician who is an expert in managing the disorder. They will work with the LMC and the whānau to make an individual plan for pēpi, which may be urgent hospital presentation for further assessment and treatment. The written report will say disorder probable.

Screened disorders that can become life-threatening in the first weeks of life include CAH, some amino acid breakdown disorders, and SCID.

If the abnormal result suggests pēpi very likely has a disorder but action is needed in days rather than hours the result will be phoned by a specialist paediatrician or a senior laboratory scientist. The result and level of urgency will be clearly explained. Further action may include referral for assessment by a local paediatrician. The report will say disorder probable.

Examples of screened disorders which are serious but not life threatening in the first weeks of life include CH, PKU, and CF.

Many disorders, such as CH and CAH, have two possible action levels for abnormal screen results, and include a borderline (slightly) abnormal level where mild disease is possible.

If the abnormal result is borderline (slightly abnormal) and suggests a disorder is possibly present, the usual next step is collection of a further blood spot specimen. The laboratory will phone the LMC to explain the result and request collection of another sample. The written report will say disorder possible.

The LMC should provide information and support to whānau who receive an abnormal screening result.

For most disorders and levels of result an information sheet is available and a link to the appropriate sheet will be texted to the LMC for information and sharing with whānau.

The laboratory can assist with further information and will ensure that LMCs are well supported when communicating abnormal results to whānau.

Each disorder in the NMSP has a specific process for diagnostic testing and treatment. Some basic information on the disorders screened for is contained in Appendix Two: Disorders in the Newborn Blood Screening Programme.

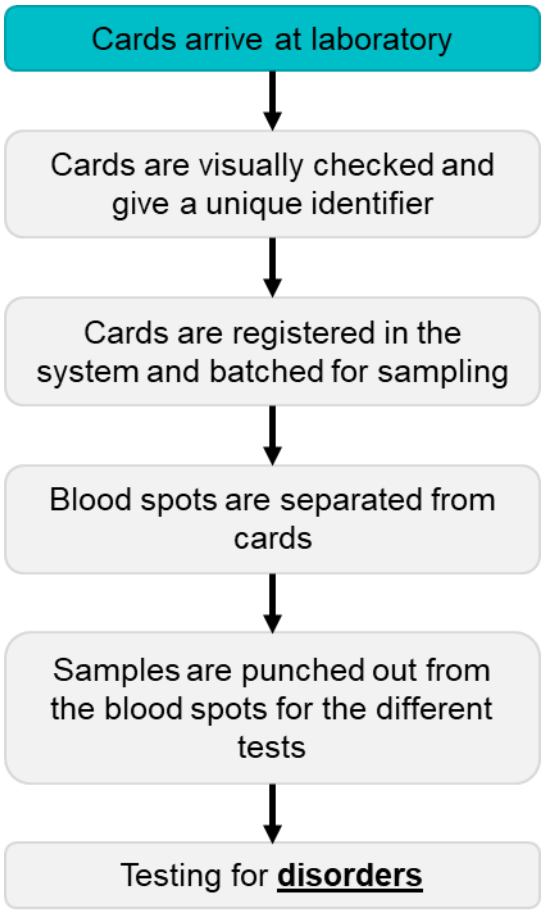
Key messages

- **LMCs must act urgently and follow directions from the laboratory or specialist paediatrician on notification of abnormal results.**
- **If LMCs are phoned by a specialist paediatrician associated with the screening programme and informed of a significantly abnormal (disorder probable) result, they will be guided by a specialist paediatrician regarding next steps.**
- **LMCs may choose to attend the paediatric consultation if the whānau wish. For metabolic disorders, assessment includes an in-person or virtual consult with the metabolic paediatrician.**
- **If LMCs receive a phone call from the laboratory advising of a slightly abnormal (disorder possible) result, they should:**
  - **contact the whānau and inform them of the screening result.**
  - **support the whānau by providing information shared by the laboratory about the reasons for a slightly abnormal result.**
  - **with parental consent, take a repeat sample and send it to the laboratory as soon as possible.**
  - **support the whānau whilst awaiting the result of the repeat sample (this will be sent by text or phone call).**
  - **notify the whānau of the results of the repeat sample.**
- **LMCs must urgently communicate significantly abnormal results to whānau with appropriate care and provide information about the disorder in a way that whānau can understand.**
- **While having regard for the need for urgency, LMCs should obtain information about the results and the disorder indicated prior to communicating results to whānau.**

11 Laboratory processes

The laboratory is responsible for testing the blood samples for metabolic and inherited disorders using the presence and levels of different markers, reporting results to LMCs, and assisting with referrals and advice for abnormal results.

When the laboratory receives blood spot cards, they are inspected for specimen quality and completeness of information then the blood portion is separated from the demographic information. The demographic information is registered into the laboratory information system and testing takes place. After testing, the cards and residual blood spots are either held indefinitely in secure storage or, on request, returned to whānau. Whānau, or individuals, can request the return of the residual blood spots at any time.



12 Data information and monitoring

12.1 Data and information collection

The NMSP collects or creates and retains the following data and information:

Pēpi	Sample	LMC/Requestor
• name • National Health Index number • sex • address • birthweight • date of birth • age at time of sample taking • gestational age at time of birth • place of birth • ethnicity • mother's name • outcome of diagnostic testing	• date and time of sample • date and time of receipt • laboratory-assigned identification number • whether this is the first sample • screening results • information about what has been reported and to whom, how and when • any clinical information that was advised to the laboratory with the sample	• name • midwifery/medical council number or common person number • contact phone numbers • contact if LMC/requestor is going to be away • preference for receiving screening reports

12.2 Uses of data and information

The NMSP securely holds the data and information it collects and retains, and uses it to:

- correctly interpret screening results
- ensure all results are given to LMCs/test requestors
- confirm that pēpi have been screened
- monitor the screening programme

Authorised personnel within the laboratory have access to the information and data for the purposes of screening and quality assurance monitoring and evaluation. Some data goes to Health NZ for monitoring reports.

LMCs must advise whānau of the data and information collected about themselves and their pēpi, and how the NMSP may use it (including for monitoring and evaluation purposes) as part of the initial discussion during pregnancy. The laboratory cannot remove information

about a screened baby once it is collected however all data is held securely and any subsequent access to this information is recorded and monitored.

12.3 Monitoring

Health NZ monitors the NMSP at a national level, through the collection of data for analysis, monitoring, and auditing. This process contributes to ongoing quality improvements.

Information related to the NMSP policy framework is at Quality standards and monitoring reports – Health New Zealand | Te Whatu Ora.

Information related to NSMP procedures and reports is at Guidelines, updates and practice reminders – Health New Zealand | Te Whatu Ora.

13 Storage, return and future use of samples

13.1 Storage or return of residual blood spots

LMCs should advise whānau that they can request the return of the residual blood spots at the time the sample is taken, or any time in the future. If they do not request their return, the residual blood spots will be stored indefinitely in secure storage and may be used for the purposes set out in Uses of residual blood spots after screening.

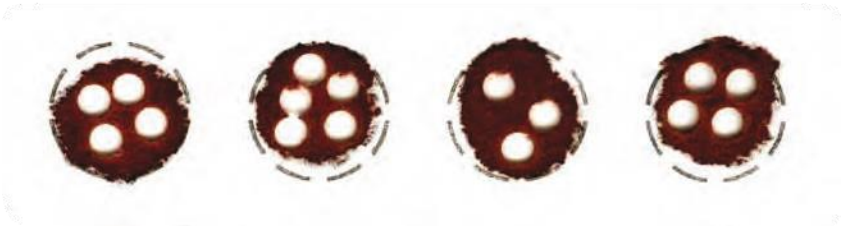
Whānau should understand the options for storage or return of residual blood spots as part of the offer of screening (see Offer of Screening). LMCs must discuss with whānau whether they want to accept or decline both:

- screening
- storage and possible future uses of the residual blood spots and explain that they can consent to screening and decline storage

The laboratory is responsible for returning residual blood spots when whānau, or individuals, request them. For samples taken after 1998 the laboratory will return the smaller portion of the card with the specimen collection paper containing the blood sample (the blood spots), but not the larger portion of the card with the demographic and other information.

The programme has protocols for secure storage and authorised access both during screening and to the residual blood sample after screening.

Figure 6: Residual blood spots after laboratory testing



13.2 How to request return of residual blood spots

Requests for return at time of sample taking
LMCs should send a request for the return, including the signature of the person entitled to request the card (eg, mother) and the address the residual blood spots are to be sent to, to the laboratory with the card. A template request form can be found at Request for return of newborn metabolic screening sample (info.health.nz).

Requests for return after sample sent to laboratory

Whānau should make a request for the return in writing using the 'Return of Newborn Metabolic Screening Samples (Guthrie Cards) to Family' form, available at Request for return of newborn metabolic screening sample (info.health.nz).

Details of who may request the residual blood spots are on the form. Photographic identification is required to ensure the laboratory sends the card to the person authorised to receive it.

The laboratory must return requested blood spots to whānau or individuals by tracked courier within one month of the request. Note that signature required courier is used so a street address must be given, not a PO box.

13.3 Uses of residual blood spots after screening

Possible uses of residual blood spots are outlined in the NMSP policy framework and on the newborn screening website.

The primary use of residual blood spots is to improve programme quality, for example through investigation of false positive and false negative results, quality assurance processes, and improving programme equipment and testing.

Additional (secondary) uses of residual blood spots may include:

- investigation of illness or deaths related to specific genetic conditions or infections at request of a healthcare practitioner and with whānau consent.
- research approved by an ethics committee towards improving the health of babies and their whānau.
- assisting the coroner / police to identify victims (of accidents, crimes, or natural disasters). The Memorandum of Understanding between the Ministry of Health | Manatū Hauora and the New Zealand Police | Ngā Pirihimana o Aotearoa can be viewed at Ministry of Health NZ.
- testing ordered by a court.

13.4 Consent for the use of residual blood spots

Consent to the use of residual blood spots to improve programme quality is part of consent to the screening process. Whānau should be advised of this during the initial discussion (see section 5.2 Initial Discussion).

The use of stored residual blood spots to investigate unexplained illness or death in a whānau only occurs when the whānau have given their informed consent. This use is for the benefit of the whānau.

Further testing on residual blood spots is only done with appropriate permission. This includes:

- from those authorised to consent (eg, whānau)
- as part of a research programme approved by an ethics committee
- through a court order.

Blood spots collected from June 2011 can be used for research that contributes to the public good, with the approval of an ethics committee and the NMSP governance group.

Note that 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. The New Zealand Code of Health and Disability Consumers' Rights 1996 allows for residual blood spots to be used without further consent from whānau or individuals for research approved by an ethics committee.

Appendix One: Contacts and resources

Sources of further information and contact details for support services are listed here.

This list should be supplemented by the local or regional services within your own networks.

Health NZ is responsible for oversight of newborn metabolic screening. Health NZ produces consumer and practitioner resources and audits and monitors this screening initiative.

Contacts

LabPLUS
Auckland City Hospital
Te Toka Tumai, Private Bag 92 024, Auckland 1142
Phone: 09 307 4949 extn 6759
Toll free: 0800 522 7587
Email: NMSP@adhb.govt.nz
http://www.labplus.co.nz

Duty Scientist, newborn screening
Ph 021 745 847

Dr Dianne Webster, Director
Email: diannew@adhb.govt.nz

Dr Natasha Heather, Lead Clinician
Email: NHeather@adhb.govt.nz

Antenatal and Newborn Screening Team
Email: Antenatalnewbornscreening@tewhatuora.govt.nz

Resources

Resources for health practitioners

- Best Practice Key Messages
- Blood Sample Protocol (Heel Prick) for Babies under 1500 grams (replaces Blood Sample Protocol for Neonatal Intensive Care Units and Special Care Baby Units)
- Heel pricks – warming, pain relief and lancet use

Consumer resource: Your newborn baby's blood test

- Hard copies are available free of charge and can be ordered at www.healthed.govt.nz or by contacting the Authorised Provider of Health Education Resources in your area. A full list of who these are (by region) is available on the HealthEd website.

On-line education for health practitioners who provide services within the antenatal and newborn screening programmes can be accessed at www.learnonline.health.nz

Fact sheets for health professional and whānau

- Congenital hypothyroidism (CH)
- Medium chain acyl-CoA dehydrogenase (MCAD) deficiency.
- Severe combined immune deficiency ()
- Cystic fibrosis (CF)
- Autosomal recessive inheritance
- Borderline positive (slightly abnormal) screen results
- Severe Combined Immune Deficiency (SCID)
- Spinal Muscular Atrophy (SMA)

Interpreter Services

Interpreter services are funded by Health NZ using Connecting Now. To access Connecting Now's interpreting service phone 0800 854 737 (PIN 14059) or email support@connectingnow.com.au

Rare Disorders New Zealand (RDNZ)

PO Box 14-313, Kilbirnie, Wellington, 6241
Phone: 0800 RARENZ (0800 727369)
Email:enquiries@raredisorders.org.nz
www.raredisorders.org.nz

RDNZ is an umbrella organisation for rare disease support groups and works with clinicians and researchers to improve health outcomes.

Genetic Services New Zealand

Northern Hub
Auckland City Hospital
Ph: (09) 307 4949 Ext. 25870
Toll Free: 0800 476 123
Email: GenSec@adhb.govt.nz

Central Hub
Wellington Hospital
Ph: (04) 385 5310
Toll free: 0508 364 436
Email: genetic.services@ccdhb.org.nz

South Island Hub
Christchurch Hospital
Ph: (03) 378 6574
Toll free: 0508 364 436
Email: genetic.servicenz@cdhb.health.nz

Health and Disability Commissioner

Office of the Privacy Commissioner

Primary Maternity Services Notice 2021

Refugee Council

WellChild/Tamariki Ora

Other resources

STAR-G disorder fact sheets

Baby's First Test

Appendix Two: The screened conditions

Summary of disorders screened for:

Disorder	Cause	Treatment	Incidence*
Congenital hypothyroidism	The thyroid is missing, not functioning or in the wrong place, which can lead to slow growth and developmental delay.	Thyroxine	1:2,500 approx. 25 babies every year
Cystic fibrosis	A defective gene and its protein product lead to thick sticky mucus.	Medication and physiotherapy	1:6,000 approx. 10 babies every year
Amino acid disorders **For example phenylketonuria (PKU)**	An enzyme is missing (in the case of PKU, lack of a particular enzyme causes an amino acid called phenylalanine to rise to harmful levels, which can lead to permanent intellectual disability and developmental delay). Other amino-acid disorders, such as MSUD, can be life-threatening.	Special diet	1:15,000 approx. 4 babies every year
Fatty acid oxidation disorders (FAODs), for example Medium chain Acyl-CoA Dehydrogenase (MCAD) deficiency	An enzyme is missing, without which energy cannot be converted from fats, which can lead to coma or death.	Regular feeding (avoidance of fasting)	1:10,000 approx. 6 babies every year
Congenital adrenal hyperplasia (CAH)	An enzyme is missing in the adrenal gland, which in severe forms can lead to ambiguous genitalia in girls, and life-threatening salt/hormonal imbalances in both sexes.	Hormone replacement	1:25,000 approx. 3 babies every year

Galactosaemia	A defective enzyme prevents normal use of milk sugar, leading to jaundice, cataracts, and life-threatening illness.	Special diet	1:110,000 approx. 1 baby every 2 years
Biotinidase deficiency	An enzyme is missing, resulting in a deficiency of biotin, which can lead to seizures, hearing loss and developmental delay.	Vitamin H (biotin)	1:180,000 approx. 1 baby every 3 years
Severe Combined Immunodeficiency (SCID)	T-cells do not develop resulting in high rate of infectious conditions	Bone marrow transplant	1:60,000 approx. 1 baby each year
Spinal Muscular Atrophy (SMA)	Motor neurons in the spinal cord are destroyed due to a defective gene	Special medicine (oral or spinal infusion)	1:10,000 approx. 6 babies every year

* For the most up-to-date incidences see Disorders screened for in the heel prick test (newborn blood spot screening) (info.health.nz)

Congenital hypothyroidism screening

Description

Congenital hypothyroidism (CH) occurs when the thyroid gland fails to develop or function properly and does not make adequate amounts of thyroxine.

Most commonly, hypothyroidism is caused by a thyroid gland that is abnormally located (ectopic), absent, underdeveloped, or severely reduced in size (hypoplastic). In these cases, the disorder is deemed to be sporadic (it occurs for no known reason).

Less commonly, the thyroid gland is present but does not produce thyroxine because of an enzyme deficiency (dyshormonogenesis). In these cases, the disorder is autosomal recessively inherited.

Thyroxine plays an important role in regulating brain development, growth, and metabolic rate. A lack of thyroxine in the first few years of life will lead to developmental delay and can lead to poor growth and short height. Other effects are low body temperature, tiredness, and constipation.

Pēpi with congenital hypothyroidism may have no obvious symptoms. However, some pēpi may be very sleepy and feed slowly. They may also have prolonged jaundice or a tendency to be constipated.

Early intervention can prevent the damage that may be caused by the disorder, and pēpi correctly treated with thyroxine can grow and develop normally.

Screening
Blood is tested for levels of thyroid stimulating hormone (TSH). Pēpi with low levels of thyroxine produce elevated levels of TSH. If the test result shows high levels of TSH, a confirmatory TSH test is performed.

There are 2 levels of abnormal screen result for CH.

- If the result is slightly abnormal and indicates that CH is possible, the laboratory will phone the LMC to request a repeat blood spot sample and will send the LMC a link to a whānau information sheet.
- If the result indicates probable CH, a specialist paediatric endocrinologist will phone the LMC and explain next steps. They will also send a link to a whānau information sheet.

Treatment
Pēpi with CH are cared for by a paediatrician (usually an endocrinologist). Treatment involves a daily oral dose of thyroxine for life (this is given to pēpi as a suspension), in sufficient quantities to bring thyroxine levels up to normal. Thyroxine levels are monitored with regular blood tests, usually weekly for six weeks then monthly until one year of age, and the thyroxine suspension is given in different amounts, depending on the blood results.

Thyroxine treatment in the correct dose is very effective and has no known adverse effects.

Cystic fibrosis screening

Description
Cystic fibrosis (CF) is an autosomal recessively inherited disorder caused by a defect in chloride transport, which causes mucus to become thick. This leads to abnormal secretions in the lungs, pancreas, intestine, liver, sweat glands and male reproductive tract. The abnormally thickened mucus in the pancreas and gut causes pancreatic insufficiency and reduced absorption of food from the gut. The lungs may be damaged by recurrent infections.

Symptoms include meconium ileus, failure to thrive and chronic respiratory problems with cough and wheeze.

Early intervention can lead to a significant improvement in quality of life and longevity. Diagnosis also enables whānau to be aware of their potential carrier status and make informed choices in the future.

Screening
Screening for CF is a two-tier process. First, blood is tested for levels of immunoreactive trypsin (IRT). This is a very non-specific measurement of pancreatic damage. Secondly, the 1 percent of blood samples with the highest levels of IRT are tested for the most common CF mutations found in Aotearoa New Zealand. There are more than 1700 possible CF mutations, but the screening programme measures only a small number. Pēpi in whom one or two mutations are present produce an abnormal screen result.

The notification to the LMC will come as a phone call from a senior scientist, advising that pēpi may either have CF or be a carrier and that further testing will determine this.

- The laboratory will send a link to a whānau information sheet.
- The arrangement for contact with the CF team and contact information will be given to the LMC at the time the result is notified.
- For possible CF, LMCs are asked to discuss the case with the CF team *before* communicating the screen result and plan to the whānau *.

Before contacting LMCs with an abnormal CF screen result, the laboratory will have checked that the local paediatric CF team is available to support the LMC and whānau. This is done to minimise the period of anxiety and distress for the whānau whilst waiting for further assessment and review with the CF team.

Pēpi who have an abnormal screening test for CF must be seen by a paediatrician for full assessment and diagnostic testing as arranged by the CF team. Diagnostic testing includes a sweat test, which measures the salt content in the baby's sweat, and further genetic testing. Pancreatic function tests may also be ordered (for example, a test for faecal chymotrypsin).

Treatment
Pēpi with CF are cared for by a clinical team which includes a CF paediatrician, physiotherapist, dietician, and social workers. Treatment involves supplementation at the earliest opportunity of pancreatic enzymes, fat soluble vitamins and salts, and special medicine may be used later.

Medications and supplements are added on an individual basis as required over time. Regular physiotherapy to treat pulmonary symptoms aims to keep the lungs as free of secretions as possible and reduce admissions associated with pulmonary exacerbations. Ongoing management of diet is required to minimise nutritional insufficiencies.

Amino acid disorders

Around 20 separate amino acids make up the building blocks of protein. Protein is usually broken down in the gut into shorter chains of amino acids and then into individual amino acids, which are absorbed into the bloodstream and processed for use in body growth and repair, making other chemicals in the body, or providing energy. Excess amino acids are broken down and excreted.

Amino acid disorders are usually the result of a deficiency in the enzymes needed to process amino acids. Unprocessed amino acids or other metabolites build up in the bloodstream and, without treatment, cause irreparable damage.

The NMSP screens for a number of amino acid disorders. Phenylketonuria (PKU) and maple syrup urine disease (MSUD) are the best known of these disorders. Further information about them is presented below.

Phenylketonuria (PKU)

Description
PKU is an autosomal recessively inherited disorder caused by lack of a liver enzyme called phenylalanine hydroxylase (or, uncommonly, a cofactor needed for its function).

Phenylalanine hydroxylase is needed to process the amino acid phenylalanine (most commonly found in protein-rich foods such as dairy products, meat, chicken, fish, eggs, nuts, beans, and lentils).

The phenylalanine which cannot be metabolised builds up in blood and tissues. This leads to toxicity and can prevent the brain from developing normally. Lack of phenylalanine hydroxylase also causes low levels of tyrosine (a neurotransmitter), which assists in the production of dopamine and serotonin.

Progressive intellectual disability occurs if PKU is not treated.

With early intervention and good treatment, pēpi with PKU can grow and develop to their full potential.

Screening
Blood is tested for levels of phenylalanine and tyrosine.

There are 2 levels of abnormal screen result for PKU.

- If the result is slightly abnormal, the laboratory will phone the LMC to request a repeat bloodspot sample and will send the LMC a link to a whānau information sheet.
- If the result indicates probable PKU a specialist metabolic physician will phone the LMC and explain next steps.

Treatment
Pēpi with PKU are cared for by a clinical team which includes a metabolic paediatrician, dietician, nurse, and local paediatrician if necessary. PKU is treated with a low-protein diet and a special nutritional supplement, both of which need careful monitoring.

Treatment is a nutritional supplement with some breastmilk or formula. The nutritional supplement contains all the essential nutrients pēpi needs, including amino acids, except phenylalanine. Phenylalanine tolerance among pēpi with PKU varies.

Regular blood tests to measure phenylalanine levels and attendance at a metabolic clinic are also part of the treatment.

Maple Syrup Urine Disease (MSUD)

Description
MSUD is an autosomal recessively inherited disorder caused by lack of an enzyme (branched-chain ketoacid dehydrogenase) that prevents the normal processing of protein and causes changes to body chemistry. The name comes from the characteristic odour of the urine of affected babies.

As a result of the deficiency, three amino acids (leucine, isoleucine, and valine) cannot be broken down, and accumulate in blood, urine, and body tissues.

In the most common severe form of MSUD, low muscle tone, lethargy, poor feeding, and hypoglycaemia develop in the first week. Without treatment, pēpi may experience seizures or coma, and may die.

Some individuals have a milder variant of MSUD and may respond to the B vitamin thiamine, but this treatment is not useful in severe forms of MSUD.

When treated from early infancy, pēpi with MSUD can grow and develop to their full potential.

Screening
Blood is tested for levels of leucine, isoleucine, and valine. If pēpi receives an abnormal screening result for MSUD, a metabolic physician associated with the screening programme will phone the LMC and explain next steps.

Treatment
Pēpi with MSUD are cared for by a clinical team which includes a metabolic paediatrician, dietician, nurse, and local paediatrician if necessary. Treatment is a low protein diet including a special nutritional supplement and some breastmilk or formula. This special diet needs careful monitoring. Regular blood tests to measure branched-chain amino acid levels and attendance at metabolic clinics are also part of the treatment.

Other amino acid disorders screened for in the NMSP

- Argininosuccinic aciduria (argininosuccinate lyase deficiency).
- Citrullinaemia (argininosuccinate synthetase deficiency).
- Glutaric acidaemia type I (glutaryl-CoA dehydrogenase deficiency).
- Homocystinuria (cystathionine beta-synthase deficiency).
- Isovaleric acidaemia (isovaleryl-CoA dehydrogenase deficiency).
- Methylmalonic acidurias (mutase deficiency, CblA, CblB, CblC, CblD defects).
- Propionic acidaemia (propionyl-CoA carboxylase deficiency).
- Tyrosinaemia Type I

All the above disorders require similar actions as PKU and MSUD. The metabolic physician and laboratory will assist with appropriate referral and answer further questions LMCs may have.

Fatty acid oxidation disorder screening

Description
Fatty acid oxidation disorders (FAODs) are autosomal recessively inherited disorders which occur because of a failure to break down fatty acids into energy. They are named according to the length of fatty acid chain that cannot be broken down. Fatty acid oxidation is the process by which a number of enzymes break down fats in the body: problems with any of these enzymes can cause a FAOD. People with FAODs cannot break down fat from either the food they eat, or fats stored in their bodies (which is the normal process when glucose from food and stored glycogen is exhausted).

Symptoms vary between different FAODs, and from person to person with the same FAOD, but hypoglycaemia is a common symptom.

Early diagnosis allows whānau to ensure that pēpi do not fast too long and have treatment regimes in place if pēpi get sick.

The NMSP screens for nine FAODs. Medium chain Acyl-CoA dehydrogenase (MCAD) deficiency is the best known of these disorders. Further information about MCAD deficiency is presented below.

MCAD deficiency

Description
MCAD deficiency occurs when the enzyme medium chain acyl-CoA dehydrogenase is either missing or not working properly within the body. This enzyme breaks down a group of fats called medium-chain fatty acids, along with fat already stored in the body, into energy.

The main source of energy for the body is glucose, obtained first from recently eaten food, then from glycogen stored in the liver, then from stored fat. Because people with MCAD deficiency cannot metabolise certain fats, their bodies cannot use fat for energy, and when they run out of glucose and glycogen, hypoglycaemia occurs. If untreated, MCAD deficiency can be fatal, but it can be easily and successfully managed if detected early.

Screening
Blood is tested for levels of acylcarnitines (products of fat metabolism). If pēpi receives an abnormal screening result for MCAD, a metabolic physician associated with the screening programme will phone the LMC and explain next steps.

Treatment
Pēpi with FAODs are cared for by a metabolic team of a paediatrician, nurse, dietitian, and social worker. They will need to attend a metabolic clinic and may need to take vitamin supplements. Treatment may be as simple as whānau advice about regular feeding and identification of symptoms. In the case of concurrent illnesses or vomiting, a care plan will include regular feeding and, if clinically required, hospital admission for nasogastric or intravenous feeding.

Other FAODs screened for in the NMSP

- Carnitine-acylcarnitine translocase deficiency (CACT).

- Carnitine palmitoyltransferase-I deficiency (CPT-I).
- Carnitine palmitoyltransferase-II deficiency (CPT-II).
- Long chain 3-hydroxy acyl-CoA dehydrogenase deficiency (LCHAD).
- Trifunctional protein deficiency (TFP).
- Multiple acyl-CoA dehydrogenase deficiency (MADD).
- Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD).

All the above disorders require similar actions as MCAD deficiency. The metabolic physician and laboratory will assist with appropriate referral and answer further questions that LMCs may have.

Congenital adrenal hyperplasia screening

Description
Congenital adrenal hyperplasia (CAH) is an autosomal recessively inherited disorder of steroid biosynthesis, usually caused by a deficiency of the enzyme 21-hydroxylase. There are other rare forms of CAH that are not screened for, and the test is unlikely to detect.

Without this enzyme the hormones used by the adrenal gland to conserve salt are not made, and the adrenal gland cannot conserve salt properly. Pēpi with CAH also have an excess of testosterone, which causes boys to reach puberty early and girls to develop masculine external genitalia.

Female pēpi with CAH may have ambiguous external genitalia at birth, but male pēpi are likely to have no obvious signs or symptoms at birth. For this reason, without screening boys in particular are at risk of adrenal crisis.

Pēpi with severe (classical or salt-losing) CAH begin to dehydrate and vomit in their first weeks of life and, if the disorder is left untreated, will die.

Early diagnosis and treatment can prevent most of the effects of the disorder.

Screening
Blood is tested for levels of 17-hydroxyprogesterone (17-OHP).

There are 2 levels of abnormal screen result for CAH.

- If the result indicates probable CAH, a specialist paediatric endocrinologist will phone the LMC and explain next steps.
- If the result is slightly abnormal and indicates that CAH is possible, the laboratory will phone the LMC to request a repeat bloodspot sample and will send the LMC a link to a whānau information sheet.

Treatment
Pēpi with CAH are cared for by a paediatrician (usually an endocrinologist). Treatment involves the replacement of cortisol to prevent the over-secretion of adrenocorticotropic

hormone (ACTH), 17-OHP and androgens, which occur in the presence of reduced cortisol levels.

Galactosaemia screening

Description
Galactosaemia is an autosomal recessively inherited disorder that prevents the normal processing of lactose (milk sugar).

Lactose is usually broken down into glucose and galactose, and the galactose is then converted to glucose for use as energy. Pēpi with galactosaemia lack an enzyme necessary to complete all the steps in this pathway. This leads to an accumulation of galactose, or metabolites of galactose, in the blood and tissues.

Without treatment, pēpi with classical galactosaemia rapidly develop hypoglycaemia, hepatic and renal damage, sepsis, and cataracts. About 80 percent of cases show distinct symptoms in the first two weeks of life, including lethargy, vomiting, jaundice, and sepsis. The remaining cases exhibit more insidious symptoms, such as vomiting, diarrhoea, failure to thrive, hepatomegaly, ascites, and cataracts.

There are several variants of the galactosaemia gene. Some mutations lead to more severe symptoms.

In some cases, a screening test may indicate abnormal galactose metabolites in a baby's blood and reduced levels of enzyme activity, but pēpi will not display any symptoms, or require treatment.

Screening may also detect a deficiency of two other enzymes involved in galactose metabolism. These are both much rarer than classical galactosaemia and cause cataracts, not acute illness.

Early intervention can prevent or reverse the acute problems of galactosaemia. However, approximately half of affected individuals may suffer some degree of learning difficulties and may have delayed speech, even when there has been no delay in diagnosis.

Screening
Blood is tested for levels of galactose metabolites (galactose and galactose-1-phosphate) and (depending on the initial results) for the most common enzyme, galactose-1-phosphate uridyl transferase (GALT).

There are 2 levels of abnormal screen result.

- If the result indicates probable galactosaemia a specialist metabolic physician will phone the LMC and explain next steps.
- If the result is slightly abnormal, the laboratory will phone the LMC to request a repeat blood spot sample and will send the LMC a link to a whānau information sheet.

Treatment
Pēpi with galactosaemia are cared for by a clinical team which includes a paediatrician, physiotherapist, dietician, and social workers. Treatment for all forms of galactosaemia is a diet without milk or milk products. Pēpi will not be able to breastfeed or drink cow's milk and will need a soy or other special milk substitute.

Treatment completely prevents or reverses only the acute problems. Some affected individuals will suffer some degree of learning difficulty and may have delayed speech, even when there has been no delay in diagnosis. Most affected girls suffer some degree of hypogonadotrophic hypogonadism (defective gonadal development or function, or failure of ovaries to develop properly).

Biotinidase deficiency screening

Description
Biotinidase deficiency is an autosomal recessively inherited disorder that prevents normal recycling of biotin (also known as vitamin H).

Biotinidase is needed to utilise biotin obtained from the diet and to reuse biotin already present in the body.

Biotin is attached to the amino acid lysine. People with biotinidase deficiency do not produce enough biotinidase enzyme to enable the body to separate the two, in order to create free biotin. Biotinidase deficiency produces a secondary deficiency of carboxylase enzymes. This can lead to irreversible neurological damage, skin disorders such as atopic or sebborrheic dermatitis and, in its most severe form, lethargy, coma and death.

Pēpi are born with biotin stores and carboxylase enzymes, and therefore symptoms do not usually appear until pēpi is several months old and the stored biotin has been depleted.

Early intervention can prevent the damage that may be caused by the disorder, and pēpi correctly treated with biotin can grow and develop normally.

Screening
Blood is tested for biotinidase enzyme. Abnormal screening results for biotinidase deficiency are re-checked by requesting another blood spot sample.

- The laboratory will phone the LMC to request a repeat blood spot sample and will send the LMC a link to a whānau information sheet.
- If pēpi receives a second abnormal screening result for biotinidase deficiency, a metabolic physician associated with the screening programme will phone the LMC and explain next steps.

Treatment
Pēpi with biotinidase deficiency are cared for by a clinical team which includes a paediatrician, physiotherapist, dietician, and social workers. Treatment involves taking biotin for life. Urine tests are used to ensure the dose of biotin is appropriate. Biotin medication is very effective, and there are no known adverse effects.

Severe combined immune deficiency (SCID)

Description
Severe combined immune deficiency (SCID) is caused by genetic defects that affect the function and development of T cells. Pēpi with SCID appear normal at birth but are born with little or no immune system. Pēpi with SCID are highly susceptible to severe infections and even common infections can be life-threatening. Without early detection and treatment, affected infants often die in the first year of life.

Screening
Blood is tested for T-cell receptor excision circles (TREC). If pēpi receives an abnormal screening result for SCID, a specialist paediatric immunologist associated with the screening programme will phone the LMC and explain next steps.

The specialist will share links to relevant information sheets.

Why does my baby need more testing?

My baby has T cells

My baby has low T cells

Treatment
The most common treatment is stem cell treatment (also called a bone marrow transplant), which uses donor cells to rebuild the immune system. Newborn screening for SCID can detect affected pēpē before they acquire infections, leading to a shorter time to transplant. Transplantation within the first 3 months of life offers the best chance for a successful outcome.

Spinal muscular atrophy (SMA)

Description
Spinal muscular atrophy (SMA) is a rare inherited neurogenetic condition. It is caused by a missing or faulty gene called the SMN1 gene. Babies usually receive two copies of this gene— one from each parent.

The SMN1 gene creates survival motor neuron protein. Without this, motor nerves in the spinal cord (motor neurons) are lost over time. Motor neurons send signals to the muscles to control muscle movement. Babies may look healthy at birth, but as motor nerves are lost, the muscles become weak, causing difficulties with motor development, breathing and swallowing.

There are several different types of SMA. SMA type 1 develops in the first 6 months of life. These babies may have breathing problems and difficulty feeding and swallowing. Without treatment this is a very severe condition which causes death in the first 2 years of life. Children with type 2 and type 3 SMA usually present after 6 months of age with low muscle tone and weakness, which affects the ability to sit or walk.

Screening

Blood is tested for the absence of any copies of the SMN1 gene. If the test result shows no SMN1 gene, a confirmatory test is performed to diagnose SMA and predict the severity of the disorder.

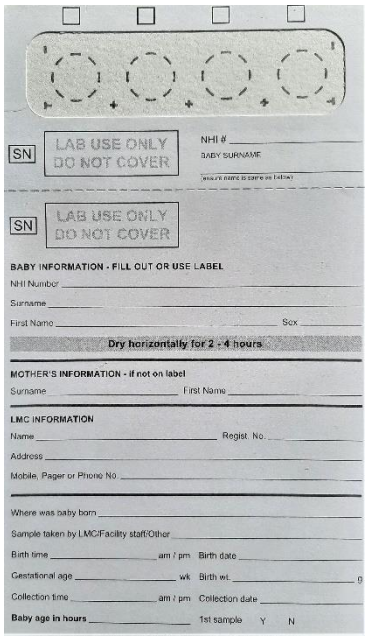
Treatment

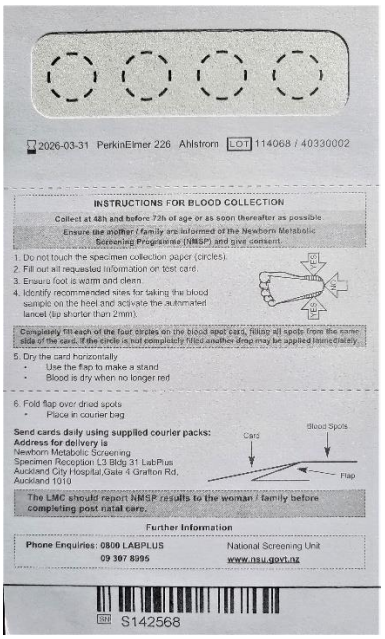
Disease modifying treatments are available for SMA. Treatment can slow and, in some cases, stop the progression of SMA, offering affected children a healthy life without significant muscle weakness.

Depending on the type of SMA, a neurologist provides detailed information about the SMA treatments available. Infants with less severe forms of SMA will be checked regularly for symptoms. Regular follow-up in a paediatric neurology clinic is important to ensure best outcomes.

Health New Zealand
Te Whatu Ora

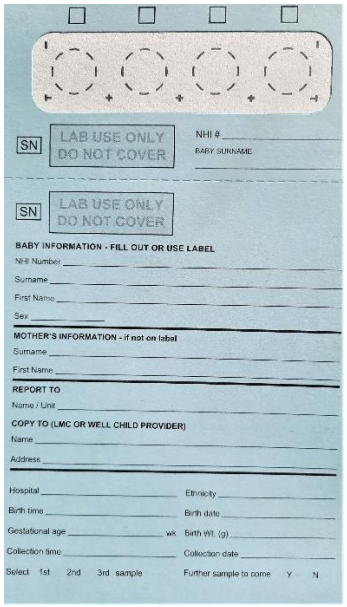
Appendix Three: Blood Spot Cards

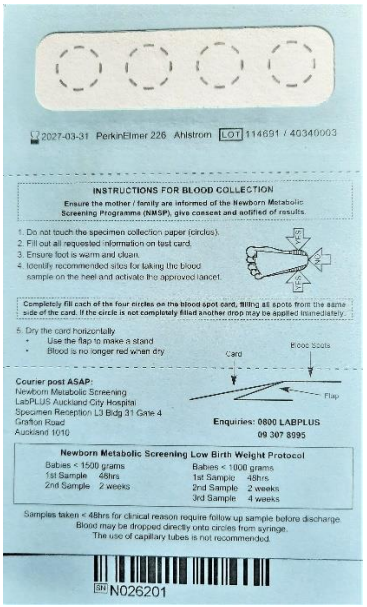




This is the standard white blood spot card used for pēpi who are born at term and are not in a Neonatal Intensive Care Unit (NICU) on the day the sample is taken.

This is the blue card used for pēpi who are born prematurely, unwell, and/or inpatients in a NICU or Special Care Baby Unit (SCBU) at the time of screening.





Appendix Four: Newborn Metabolic Screening Protocol for Premature Babies

Preterm Metabolic Bloodspot Screening Protocol (from 1 July 2024)

Scope of the protocol

This protocol describes the timing of screening for newborns in NICU or Special Care Baby Unit (SCBU).

Overview of the screening protocol

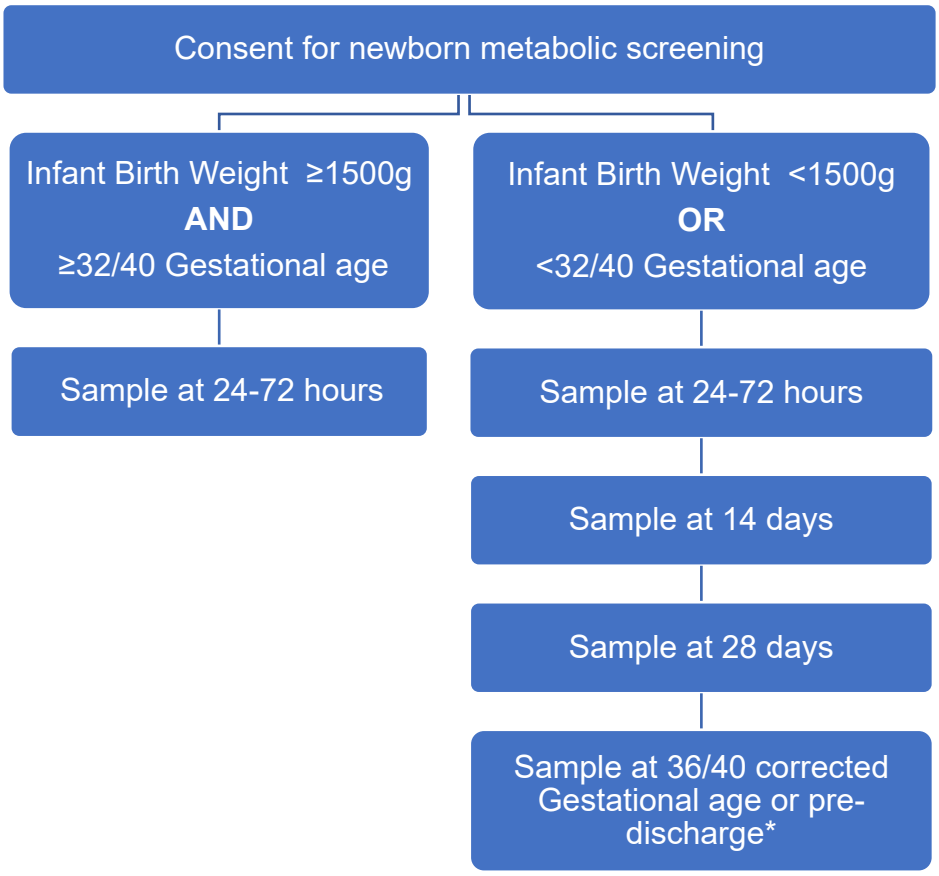
The newborn metabolic screening protocol in Aotearoa New Zealand is to obtain a blood spot sample from all babies from 24 hours of age once informed consent is obtained from the whānau for collecting and storing or returning the sample.

Many babies that are born preterm, have very low birth weight, or are sick have false positive screens for screened conditions at 24 hours. There is also a risk of screening missing the diagnosis of congenital hypothyroidism because the pituitary hypothalamic axis is insufficiently developed to produce an elevated level of TSH in response to a low thyroxine level. Thus, a preterm protocol is needed to ensure that these babies are appropriately screened for congenital hypothyroidism and other disorders.

General principles

- Blue coloured blood spot cards are to be used for all babies in a NICU or SCBU at the time of screening, or who are under 32/40 gestational age or 1500g birthweight.
- Results highly suggestive of a screened disorder will be phoned.
- All results will be reported.
- Reporting will be to the individual(s) and/or role named on the card.

PROTOCOL FOR ROUTINE SAMPLES:



*Unless there has already been a bloodspot sample collected within 2 weeks.

Additional key points:

- The screening programme does not send reminders that further samples need collecting.
- Complete screening and correct result interpretation and reporting rely on clinical staff filling the forms correctly and taking the samples at the right time.
- For further details on the Newborn Metabolic Screening Programme (NMSP) please see Newborn Metabolic Screening Programme – Health New Zealand | Te Whatu Ora.
- For screening results please call LabLINK at Auckland District Health Board on 0800 522 7587.

This protocol has been developed in consultation with Aotearoa New Zealand endocrinologists, general paediatricians and neonatologists.

This protocol replaces the Newborn Metabolic Screening Protocol for Babies Under 1500 grams.

Any queries regarding this protocol, please call the screening laboratory Duty Scientist on 021 974 5847.

Appendix Five: Newborn metabolic screening protocol for babies who have received blood transfusions

Scope of the protocol

This protocol describes screening for specific metabolic and inborn disorders in newborns who have received blood transfusions.

Overview of the screening protocol

Metabolic screening protocol in Aotearoa New Zealand to date is to obtain a heel prick sample from all pēpi from 24 hours age when consent has been obtained from the whānau. Blood transfusions may affect the results of the screening tests.

General principles

- A pre-blood transfusion sample should be taken if possible
- A sample should be taken at 24 - 72 hours of age regardless of whether an earlier sample was taken
- If a pre-blood transfusion sample was not taken, another sample is required at three months after the last blood transfusion
- The date of the last blood transfusion must be recorded on the blood spot cards

Note: *the screening programme will no longer be sending reminders that further samples need collecting – complete screening and correct result interpretation and reporting rely on clinical staff filling the forms correctly and taking the samples at the right time.*

For further details on the Newborn Metabolic Screening Programme (NMSP) please see Newborn Metabolic Screening Programme – Health New Zealand | Te Whatu Ora.

Any queries regarding this protocol, please call the screening laboratory Duty Scientist on 021 974 5847.