National Screening Unit

Guidelines for practitioners providing services within the Newborn Metabolic Screening Programme in New Zealand

February 2010

New Zealand Government

Purpose of the Newborn Metabolic Screening Programme

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 disorders in pre-symptomatic babies. This service is offered to all babies born in New Zealand.

The NMSP is one of the most successful screening programmes in New Zealand, and has resulted in the diagnosis of more than 900 babies with metabolic disorders since its commencement in 1969. For babies identified as being at increased risk, and their families, the benefits of screening are enormous. Early treatment can improve babies' health and prevent severe disability and death.

Acknowledgements

These Guidelines have been developed in consultation with the Newborn Metabolic Screening Programme Advisory Group.

The National Screening Unit thanks the Advisory Group and the many individuals and groups who contributed feedback to drafts of this document.

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MINISTRY OF HEALTH
MANATŪ HAUORA

CONTENTS

List of acronyms

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

Definitions

In these Guidelines "Lead Maternity Carer" (LMC) has the same meaning as in the Primary Maternity Services Notice 2007:

Lead Maternity Carer (LMC) means a person who:

- is a general practitioner with a Diploma in Obstetrics (or equivalent, as determined by the New Zealand College of General Practitioners), or a midwife, or an obstetrician; and
- is either a maternity provider in his or her own right, or an employee or contractor of a maternity provider; and
- has been selected by a woman to provide her lead maternity care.

Mā te rongo, ka mōhio;
Mā te mōhio, ka mārama;
Mā te mārama, ka mātau;
Mā te mātau, ka ora.

Through feeling comes awareness;
through awareness comes understanding;
through understanding comes knowledge;
through knowledge comes life and wellbeing.

Key messages

1. The success of the NMSP depends on the diligence and dedication of many health professionals, and LMCs in particular. Their input is integral to the continuation of a high-quality screening programme. Page 1

2. Many of the disorders the NMSP screens for can lead to serious illness or mortality within seven to ten days. Samples must be taken at 48 hours after birth or as soon as possible after this to prevent irreversible damage and life-threatening illnesses. Page 3

3. LMCs are responsible for the newborn metabolic screening process, including giving information and advice, offering screening, ensuring informed consent, documenting the process, taking a suitable sample and following up results. Page 8

4. Appropriate information about the NMSP must be given to parents/guardians during pregnancy. Page 9

5. Clear documentation of the screening process must be kept, including consents, declines, and sample information. Page 11

6. Samples must be taken at 48 hours after birth or as soon as possible after this. Page 16

7. Samples must be suitable for testing. Page 19

8. Samples must be sent to the laboratory as soon as they are dry. Page 23

9. Requests for repeat samples must be acted on urgently. Page 24

10. If there are clinical concerns about a baby, the baby must be referred to diagnostic and treatment services without waiting for screening results. Page 27

11. LMCs must reconcile laboratory reports with samples they have sent for testing, and clarify any discrepancies with the laboratory. Page 27

12. If a baby receives a positive screening result, LMCs must take further action, according to the advice of laboratory staff. Page 29

13. Parents/guardians can request that residual blood spots be returned after screening. Page 35

1 INTRODUCTION

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

These Guidelines for best practice are to support practitioners offering services within the Newborn Metabolic Screening Programme (NMSP). The Guidelines are for all practitioners involved in aspects of the NMSP, including Lead Maternity Carers (LMCs), hospital midwives, nurses and phlebotomists.

LMCs have a contractual obligation under the Primary Maternity Services Notice 2007, issued pursuant to section 88 of the New Zealand Public Health and Disability Act 2000, to provide services within screening programmes endorsed by the Ministry of Health, including the NMSP.

LMC responsibilities include:

- providing appropriate information and education about screening
- offering screening
- ensuring screening is performed
- meeting programme standards
- ensuring any follow-up actions requested by the screening programme are completed.

These Guidelines should be read in conjunction with the NMSP standards, policy documentation, and education resources produced by the National Screening Unit (NSU) and available on the NSU's website at www.nsu.govt.nz

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

2 BACKGROUND INFORMATION

2.1 Overview

The NMSP is one of the most successful screening programmes in New Zealand. Almost all babies born in New Zealand are screened, and as a result approximately 45 babies are identified with and treated for a metabolic disorder each year. When metabolic disorders are diagnosed in early infancy, treatment can commence immediately, preventing irreversible damage and life-threatening illnesses.

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

There are hundreds of metabolic disorders, many of which have only recently been discovered. The NMSP screens for a small percentage of these disorders, focusing on those for which appropriate testing is available and that can be successfully treated in the early newborn period, for the benefit of affected babies and their families.

The NSU has responsibility for the funding, monitoring and strategic direction of the NMSP. The programme is currently run by the Auckland District Health Board laboratory (LabPLUS) at Auckland City Hospital.

2.2 A history of newborn metabolic screening in New Zealand

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. By 1978, assays were sufficiently sensitive to test for congenital hypothyroidism (CH). In 1979, New Zealand scientists 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. Congential 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 7 to 28 metabolic disorders, with the addition of 9 fatty acid oxidation disorders (FAODs) and 12 more amino acid disorders.

2.3 Disorders in the Newborn Metabolic Screening Programme

The 28 metabolic disorders currently screened for by the NMSP are:

- Amino acid disorders (14 disorders, including PKU and MSUD)
- Fatty acid oxidation disorders (9 disorders)
- Others:
  - Congenital hypothyroidism
  - Cystic fibrosis
  - Congenital adrenal hyperplasia
  - Galactosaemia
  - Biotinidase deficiency.

For more information on these disorders see Appendix Two: Disorders in the Newborn Metabolic Screening Programme.

2.4 Potential benefits and harms of newborn metabolic screening

Many of the disorders the NMSP screens for can lead to serious illness or mortality within seven to ten days. Samples must be taken at 48 hours after birth or as soon as possible after this to prevent irreversible damage and life-threatening illnesses.

The NMSP provides the greatest benefit to babies and families of babies who receive a positive screening result. Early diagnosis and treatment can allow babies with metabolic disorders to reach their full potential. Minimising the morbidity caused by metabolic disorders also has benefits at a society-wide level, reducing costs to health, disability and education services.

Societal benefits of positive screening results include:

- reduced cost in managing the health of individuals with morbidity associated with metabolic disorders (offset by the comparatively low cost of the screening programme)
- reduced cost of long-term social support for individuals with metabolic disorders
- raised awareness of metabolic disorders in the health sector and the community
- potential future benefits from the use of residual blood spots, which are currently stored on a long-term basis.[1]

The majority of screened babies will receive a negative result. This can reassure families that their baby is unlikely to have one of the metabolic disorders screened for.

Potential harms of the NMSP are:

- that screening is not diagnostic and there is the possibility of false positive and false negative results
- the possibility that practitioners may ignore clinical symptoms caused by a disorder if a screening result is negative
- parental anxiety associated with waiting for results
- that parents/guardians may receive insufficient and/or inappropriate information to allow them to make an informed choice about screening
- for babies with positive results:
  - morbidity associated with delayed diagnosis if samples are unsuitable, taken late or delayed in transit to the laboratory
  - inequities associated with potential lack of access to specialist metabolic services.

These are all potential harms that can be ameliorated by high-quality service provision, health practitioner education and effective monitoring of the NMSP.

[1]The Ministry of Health is currently reviewing policies regarding the storage, retention, and uses of residual blood spots. Any changes to policies will be notified on the NSU website (www.nsu.govt.nz) and reflected in the online version of these Guidelines.

3 GENERAL PROGRAMME REQUIREMENTS

3.1 Code of Health and Disability Services Consumers' Rights

The Code of Health and Disability Services Consumers' Rights provides that New Zealand health care consumers have a legal right to appropriate information to enable them to give informed consent. Information about the Code can be obtained from the Health and Disability Commissioner's website, www.hdc.org.nz

3.2 Health Information Privacy Code

The Health Information Privacy Code 1994 (HIPC) sets specific rules for agencies in the health sector to better ensure the protection of individual privacy. It addresses health information collected, used, held and disclosed by health agencies.

For the health sector the HIPC takes the place of the information privacy principles set out in the Privacy Act 1993. The HIPC can be viewed at the Privacy Commissioner's website, www.privacy.org.nz

3.3 Tailoring services to individual needs

Health services should be tailored to meet the needs of the individuals receiving them. This helps to ensure equity of access and outcomes. Guidance on how to achieve this is set out in the Ministry of Health's Statement of Intent for 2009 – 2012. The table below highlights some key aspects of the Statement of Intent.

Achieving equitable outcomes	
GUIDELINE	**INFORMATION**
The Ministry of Health promotes evidence-based and cost-effective service models which emphasise early intervention, including prevention and self-management. Health services should: • enable people to take responsibility for managing their own health, make healthy lifestyle choices, and progress Whānau Ora using the Whānau Ora tool, available at www.moh.govt.nz • improve early intervention in childhood to reduce the likelihood of minor child health problems becoming major adult health problems.	LMCs should incorporate these principles into their own practices and tailor services to meet the individual needs of parents/guardians.
Health services should be attuned to the needs of patients, individuals, families, and communities.	Health services should be tailored to meet the health needs of all New Zealanders, including Māori, Pacific peoples and Asian populations. Health providers should recognise that what works for different populations varies.

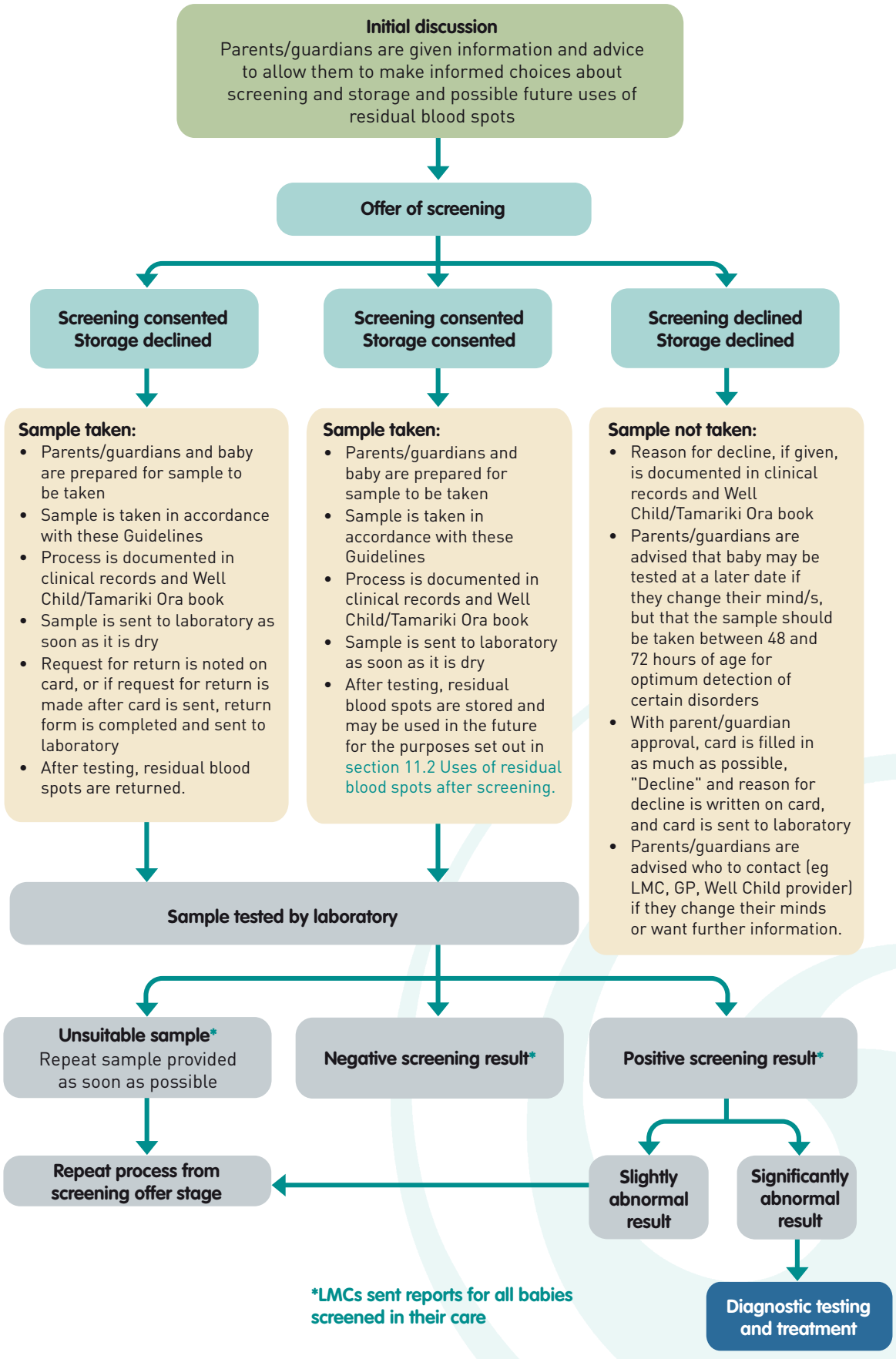
Whānau Ora	
GUIDELINE	**INFORMATION**
Whānau Ora is about: • facilitating positive and adaptive relationships within whānau • recognising the interconnectedness of health, education, housing, justice, welfare, employment, and lifestyle as elements of whānau wellbeing • ensuring Māori families are supported to achieve their maximum health and wellbeing.	LMCs should familiarise themselves with the Whānau Ora tool, which is available on the Ministry of Health website at www.moh.govt.nz

3.4 Use of interpreter services

LMCs may need to use interpreter services to ensure that parents/guardians understand the information provided to them about newborn metabolic screening. Using friends or family members as interpreters is not recommended practice.

Using interpreter services	
GUIDELINE	**INFORMATION**
LMCs should use interpreting services to aid understanding, if appropriate.	LMCs may arrange an interpreter: • if requested by the parents/guardians • if parents/guardians have a limited command of English • if they are concerned that the parents/guardians do not understand the clinical information • where a parent/guardian is deaf and understands sign language. When determining whether to use an interpreter, LMCs may consider whether the following considerations are being met: • the principles of informed consent • the need for effective communication • confidentiality • ensuring the best patient outcome.

4 THE NEWBORN METABOLIC SCREENING PROCESS



5 SPECIFIC PROGRAMME REQUIREMENTS

5.1 Responsibility for newborn metabolic screening

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

LMCs are responsible for newborn metabolic screening. If a woman is receiving care away from her LMC, aspects of screening may be undertaken by the primary maternity carer or health provider at the time.

If care has been transferred and the woman is in the care of the secondary/tertiary service, that service is responsible for the screening process.

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

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

5.2 The screening process

The screening process starts with the provision of information to parents/guardians 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 the baby screened receives a negative result or is referred for diagnostic testing, or if the parents/guardians withdraw from the process.

When laboratory testing is complete, the residual blood spots are either held in secure storage indefinitely or returned to parents/guardians (see Chapter 11: Storage, return, and future uses of residual blood spots). Stored blood spots may be used in the future for the purposes listed in section 11.2: Uses of residual blood spots after screening.[2] If parents/guardians or the individual want the blood spots returned from storage, they must make a written request.

5.3 Family history of disorders

If any of the metabolic disorders screened for are present in a baby's family, the baby may have an increased chance of inheriting the disorder. It may be appropriate for the baby to undergo diagnostic testing as well as being screened. LMCs should refer parents/guardians with a family history of disorders to a paediatrician for further advice during pregnancy.

[2]The Ministry of Health is currently reviewing policies regarding the storage, retention and uses of residual blood spots. Any changes to policies will be notified on the NSU website (www.nsu.govt.nz) and reflected in the online version of these Guidelines.

6 INFORMED CONSENT

6.1 Informed consent requirements

Ensuring that parents/guardians give informed consent for newborn metabolic screening is a legal requirement that is central to best practice in maternity care.

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 uses of blood spot cards.

These points are covered in more detail in the following sections.

6.2 Provision of information to parents/guardians

Appropriate information about the NMSP must be given to parents/guardians during pregnancy.

Parents/guardians should be provided with information about the NMSP and asked to consider having their baby screened, during pregnancy. Further information should be provided to parents/guardians as required after the birth.

The information given to parents/guardians should be tailored to each step in the screening pathway and to the specific needs of the parents/guardians.

Information and resources to assist with this process are listed in Appendix One: Sources of support and further information.

Provision of information	
GUIDELINE	**INFORMATION**
LMCs must provide information in a way that parents/guardians can understand.	Information should include: • discussions between the LMC and parents/guardians throughout antenatal care • the newborn metabolic screening consumer pamphlet • viewing of the newborn metabolic screening consumer DVD • referral to other sources of information, such as websites and specialist organisations. Information should be communicated with regard to individual levels of understanding and background knowledge.

Additional support	
GUIDELINE	**INFORMATION**
LMCs must offer additional support to parents/guardians who have difficulty understanding information because of language difficulties, hearing impairment or intellectual disability.	Appropriate information that allows for informed consent includes using professional interpreter services, such as Language Line, a DHB interpreter or a New Zealand sign language interpreter, where necessary. Using family members or friends as interpreters is not recommended practice (see section 3.4: Use of interpreter services). Parents/guardians who have an intellectual disability may require extra support or the presence of family members or other support people to understand the information.

Questions	
GUIDELINE	**INFORMATION**
LMCs must answer questions parents/guardians ask about the NMSP.	Parents/guardians should be given the opportunity to ask questions about the NMSP, and advised where they can find further information. LMCs may seek advice from the NMSP to assist them to answer questions from parents/guardians.

6.3 Documentation

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

Written consent for newborn metabolic 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 LMCs, or if there is no LMC assigned, the antenatal care provider.

Clinical records	
GUIDELINE	**INFORMATION**
LMCs must keep a record of the screening process.	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 positive 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.

6.4 Initial discussion

LMCs must initiate a discussion with parents/guardians about the NMSP during pregnancy, to give them the opportunity to consider participation in the NMSP, ask questions and seek further information.

The discussion should take into account any barriers to understanding (such as language or disability), and parents/guardians should be advised that family members may be present at the discussion, if they wish.

Parents/guardians, on behalf of the baby, should understand the purpose, benefits and potential harms of participating in the NMSP, and know that they can withdraw from the process at any time before the sample has been taken.

The discussion must include the following information.

1. **About the NMSP**
   - The purpose of screening.
   - The disorders screened for.
   - How screening can help babies who have metabolic disorders.
   - That screening does not cover every 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.

2. **Resources**
   - Availability of the DVD and other consumer resources.
   - Availability of further information.

3. **Benefits and harms of screening**
   - That the disorders screened for cannot be easily detected without blood tests.
   - That the disorders screened for can be life-threatening.
   - That early detection of the disorders screened for can enable early treatment.
   - That screening is not diagnostic, and there is a possibility of false positive and false negative results.
   - The benefits and risks associated with screening.

4. **Consent**
   - That screening is voluntary.
   - That parents/guardians may decline screening on the baby's behalf.
   - That if a screening result is positive, diagnostic testing will then be required, and referral for diagnostic testing may be made without further consent being sought.

- That consent is required both for screening and for storage and possible future uses of residual blood spots.
- The right of parents/guardians to withdraw consent if they change their minds before the sample has been taken.

5. Sample taking

- How the blood sample is taken.
- How parents/guardians can prepare their baby for and comfort their baby during sample taking.
- That a repeat sample is needed for about one in every 50 babies.

6. Results

- That all results are notified to LMCs.
- That unsuitable samples, slightly abnormal results, CF and biotinidase deficiency results are notified by letter to the LMC within ten days of the sample reaching the laboratory.
- That significantly abnormal results and results that suggest urgent clinical intervention is required are notified to the LMC by phone, and may have to be acted on urgently.

7. Data and information collection and monitoring

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

8. Storage, return, and future uses of residual blood spots

- That screening can be undertaken without storage and possible future uses (ie, residual blood spots can be returned to the family/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 parent/guardian, or the individual, to request return of the residual blood spots at any time.

6.5 Offer of screening

LMCs are responsible for offering screening. If care has been transferred or there is no LMC assigned for maternity care, the antenatal care provider at the time is responsible for offering screening.

Parents/guardians must decide:

- whether they agree to their baby being screened for metabolic disorders
- if they agree to screening, whether they agree to their baby's residual blood spots being stored for possible future uses, or want the residual blood spots returned after screening.

LMCs must make it clear to parents/guardians that they have two separate decisions to make, and facilitate informed choices for each.

Offering screening	
GUIDELINE	**INFORMATION**
LMCs must offer all parents/guardians newborn metabolic screening for their babies.	All parents/guardians must be offered newborn metabolic screening for their babies with sufficient information, advice, and time to enable them to make an informed decision.

6.6 When screening is declined

The Ministry of Health strongly recommends that all babies are screened.

However, parents/guardians have the right to accept or decline the screening offer. If parents/guardians decline screening, their decision is usually advised to the LMC during pregnancy. LMCs should document the decision in the clinical records and handover notes to Well Child/Tamariki Ora providers and other health care providers.

When screening is declined	
GUIDELINE	**INFORMATION**
LMCs must inform parents/guardians of their right to decline the screening offer on behalf of their baby.	Parents/guardians have the right to accept or decline the screening offer, and to choose whether to have their baby's residual blood spots stored for possible future uses, or returned.
LMCs must provide parents/guardians with sufficient information and advice to enable them to understand the implications of declining to participate in the NMSP.	If parents/guardians decline to have their baby screened, LMCs should be satisfied that the decision has been made on the basis of appropriate information and advice. While respecting the parents/guardians' decision, LMCs may wish to check that parents/guardians have been provided with the Ministry of Health newborn metabolic screening consumer resource, are aware of the purpose of screening, and have been given information and advice about the NMSP. Parents/guardians should be: • advised that screening can be undertaken without storage and possible future uses of residual blood spots • advised that if they change their minds the sample can be taken later, but that this may not be the optimum time for effective screening • offered further information, if appropriate.
LMCs must document when screening is declined, and the reason, if one is given, in clinical records and handover notes to Well Child/Tamariki Ora providers and/or GPs and primary carers.	
LMCs should ask parents/guardians if they agree to a blood spot card being filled out with their demographic information, to monitor participation in the NMSP.	If parents/guardians agree, the card should be filled in with the baby's demographic information, LMC details, and a note that screening was declined, and sent to the laboratory.

7 SAMPLE TAKING

7.1 Responsibility for taking samples

LMCs are responsible for ensuring samples are taken if consent is given.

If care has been transferred and the woman is in the care of the secondary/tertiary service, that service is responsible for ensuring a sample is taken if consented.

If there is no LMC assigned for maternity care, the antenatal care provider or the secondary/tertiary service is responsible for ensuring a sample is taken.

7.2 Ensuring informed consent

If the sampling practitioner is not the person who made the screening offer, they must:

- inform the parents/guardians that they have a request form for taking a blood sample
- confirm that the parents/guardians have agreed to have the sample taken.

If parents/guardians 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 metabolic screening, the sampling practitioner should not take the sample, and inform the LMC or antenatal care provider as soon as possible that the sample has not been taken, and the reason.

The LMC or antenatal care provider should:

- discuss the NMSP with parents/guardians and provide information and advice in accordance with section 6.2: Provision of information to parents/guardians and section 6.4: Initial discussion of these Guidelines
- document the process in the clinical records.

7.3 Timing for taking samples

Samples must be taken at 48 hours after birth or as soon as possible after this.

Taking samples when the baby is 48 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 metabolic disorders screened for can be fatal within seven to ten days, but may not show any signs or symptoms until irreversible damage has occurred. In many metabolic disorders, biochemical levels are normal at birth because, while the baby was *in utero*, the placenta eliminated abnormal biochemicals as the baby's system produced them. For this reason, babies affected by a disorder can be born without any signs or symptoms.

For effective newborn metabolic screening, the baby must have been independent from the mother long enough for the indicator biochemicals to show an abnormality. In babies with FAODs, the catabolic state all babies experience (when they lose birth weight because they are not feeding sufficiently to meet their body needs) changes the indicator biochemicals. The fatty acid biochemicals normalise when the baby starts feeding well. In other disorders (for example, amino acidopathies) the biochemicals 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. Therefore the optimum window of opportunity is between 48 and 72 hours.

Timing for taking samples	
GUIDELINE	**INFORMATION**
LMCs must ensure that samples are taken as soon as possible after the baby is 48 hours old and prior to 72 hours old, to prevent irreversible damage and life-threatening illnesses.	It is possible for samples to be taken later if parents/guardians initially decline screening but then change their minds, even though the optimum time for sample-taking has passed. Milk feeding, stool colour or antibiotic use does not affect results, and must not delay sample-taking.

7.4 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.

Blood spot card information	
GUIDELINE	**INFORMATION**
Blood spot cards must be completed with all requested information.	All information requested on the blood spot card is needed by the laboratory to ensure a high quality screening test. Screening results may be inaccurate if blood spot cards are not correctly filled out.

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

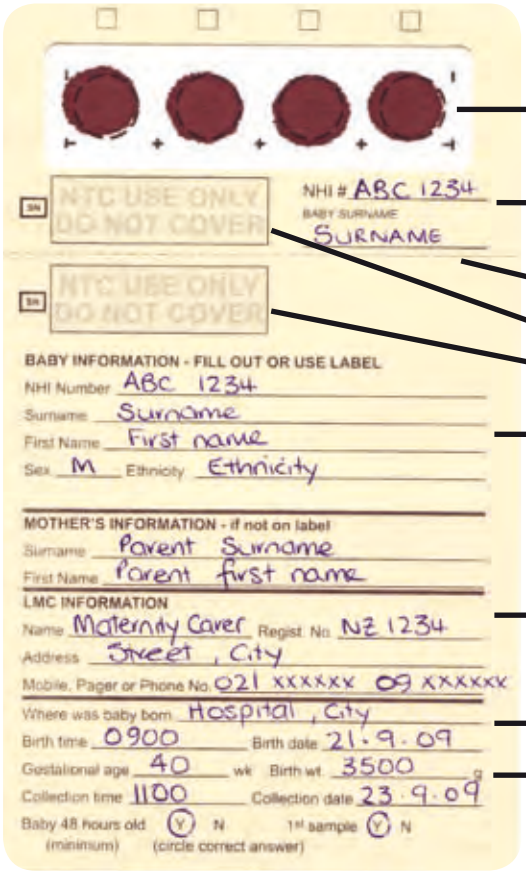
LMC details	
GUIDELINE	**INFORMATION**
LMC contact details must be provided on the blood spot card, including a contact telephone number.	Include details for a back-up contact if the LMC is going to be away.

Figure 1: Blood spot card

Please fill in all fields on the card.

Do not use staples or sellotape on the card.

Post the card to the lab as soon as it is dry (1 ½ - 2 hours).



Specimen collection paper.

Written information only (no stickers).

Perforation for automatic punching.

Please do not put stickers over the NTC areas.

Written information or hospital sticker.

The LMC is the point of contact for positive results. Give alternative contacts if you will be away.

Critical for testing and interpreting results.

Sample must be taken at 48 hours of age or as soon as possible thereafter.

7.5 Procedure for taking samples

Samples must be suitable for testing.

Figure 2: Correct sample-taking procedure

Note that protocols are in use for babies admitted to neonatal intensive care units (NICUs) and special care baby units (SCBUs), (see Appendix Four: SCBU and NICU Screening Protocols).

1. Obtain informed consent from parents/guardians.

Obtain consent for:

- screening
- storage and possible future uses of residual blood spots (see Chapter 11: 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 is provided in case the laboratory requires an urgent contact for positive results.

3. Gather equipment.

Equipment needed for the procedure is:

- gloves
- cleansing material (if required)
- approved lancet (see step 9)
- completed blood spot card.

4. Ensure baby is warm and comfortable.

Analgesia in the form of breastfeeding, bottle feeding or skin-to-skin contact is beneficial.[3] Encourage the baby to breastfeed or be cuddled during the procedure.

[3]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.

5. **If there is any need to warm the baby's foot, do so with care.**

   A warm heel is needed for good perfusion. Warm with booties and blankets if necessary.[4]

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 the baby from infection and the sampling practitioner from blood contamination.

7. **Make sure the baby's foot is clean and dry.**

   If necessary, clean the baby's foot with warm water or an alcohol swab, and allow to dry before taking the blood sample.

8. **Allow the baby's 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 babies.

   There are a number of approved lancets available in New Zealand. 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 on the NSU website, www.nsu.govt.nz

Figure 3: Quikheel™ lancet and Tenderfoot® lancet

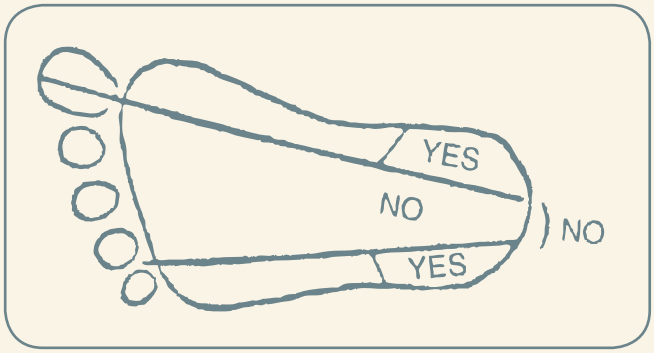
[4]NMSP. 2008. Heel pricks – warming, pain relief and lancet use.

Various studies have examined the effectiveness of different lancet types ('stabbers' versus 'slicers'). As a result the NSU recommends the slicing device, which generally decreases the need for more than one heel prick per test and reduces crying and pain for the baby.[5]

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

To avoid calcaneal puncture and the risk of osteochondritis, 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 the spot is filled.
- 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

[5]NMSP. 2008. Heel pricks – warming, pain relief and lancet use.

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 section 7.6: Sending samples to laboratory.

Satisfactory sample collection	
GUIDELINE	**INFORMATION**
LMCs must ensure that samples are taken, dried and sent in accordance with these Guidelines.	If samples are unsuitable (see section 9.3: Unsuitable samples), a repeat sample will be needed and screening will be delayed. If the baby has one of the disorders screened for, this delay could compromise the baby's access to early diagnosis and treatment.

7.6 Sending samples to laboratory

Samples must be sent to the laboratory as soon as they are dry.

Samples must never be batched over several days for posting, even if they are being sent from a hospital.

Samples must be sent to the laboratory 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.

Cards sent via a hospital's internal mailing system may not reach the laboratory as quickly as those sent via New Zealand Post.

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.

The sample-taking and sending must be recorded in the clinical notes.

7.7 Use of capillary tubes and arterial lines

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

Capillary tubes with anticoagulent must not be used.

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

7.8 Low birth weight and sick babies, and effect of blood transfusions

Low birth weight and sick babies often receive false positive results due to their immaturity. Blood transfusions can also affect test results. Protocols have been developed to ensure effective screening for these babies (see Appendix Four: SCBU and NICU Screening Protocols).

7.9 Repeat samples

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 positive results. For more information about unsuitable samples and positive results, see Chapter 9: Screening results.

The laboratory will advise the LMC why the sample was unsuitable or what the positive results indicated.

If the result indicates a severe form of one of the disorders, the laboratory will telephone the LMC, request an urgent repeat sample, and advise the LMC of any referral or follow-up needed.

If the result indicates a mild form of one of the disorders, the laboratory will send a letter to the LMC requesting a repeat sample and providing information about the indicated disorder.

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

If the sample was unsuitable, the laboratory will send a letter to the LMC requesting a repeat sample, advising that the sample was unsuitable and could not be fully tested, and the reasons for this.

LMCs must follow the repeat sample procedure below, having regard to the level of urgency advised by the laboratory.

Repeat samples	
GUIDELINE	**INFORMATION**
LMCs must provide parents/guardians with sufficient information about repeat samples.	A repeat sample is required for about one in every 50 babies. Repeat samples are required for unsuitable samples and positive results. In both of these cases, LMCs should offer further information and guidance to parents/guardians.

7.10 Repeat sample procedure

1. Contact the parents/guardians.

- Contact the parents/guardians and explain why a repeat sample is necessary.
- If sample result/s were positive, advise the parents/guardians of the result/s.
- If the baby is unwell, refer to a GP or paediatrician.

If the LMC is unable to contact the parents/guardians:

- the laboratory must be notified
- the mother's GP or Well Child/Tamariki Ora provider should be notified that the laboratory has requested a repeat sample.

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.

3. Take the repeat sample in accordance with Guidelines for the first sample.
(see section 7.5: Procedure for taking samples).

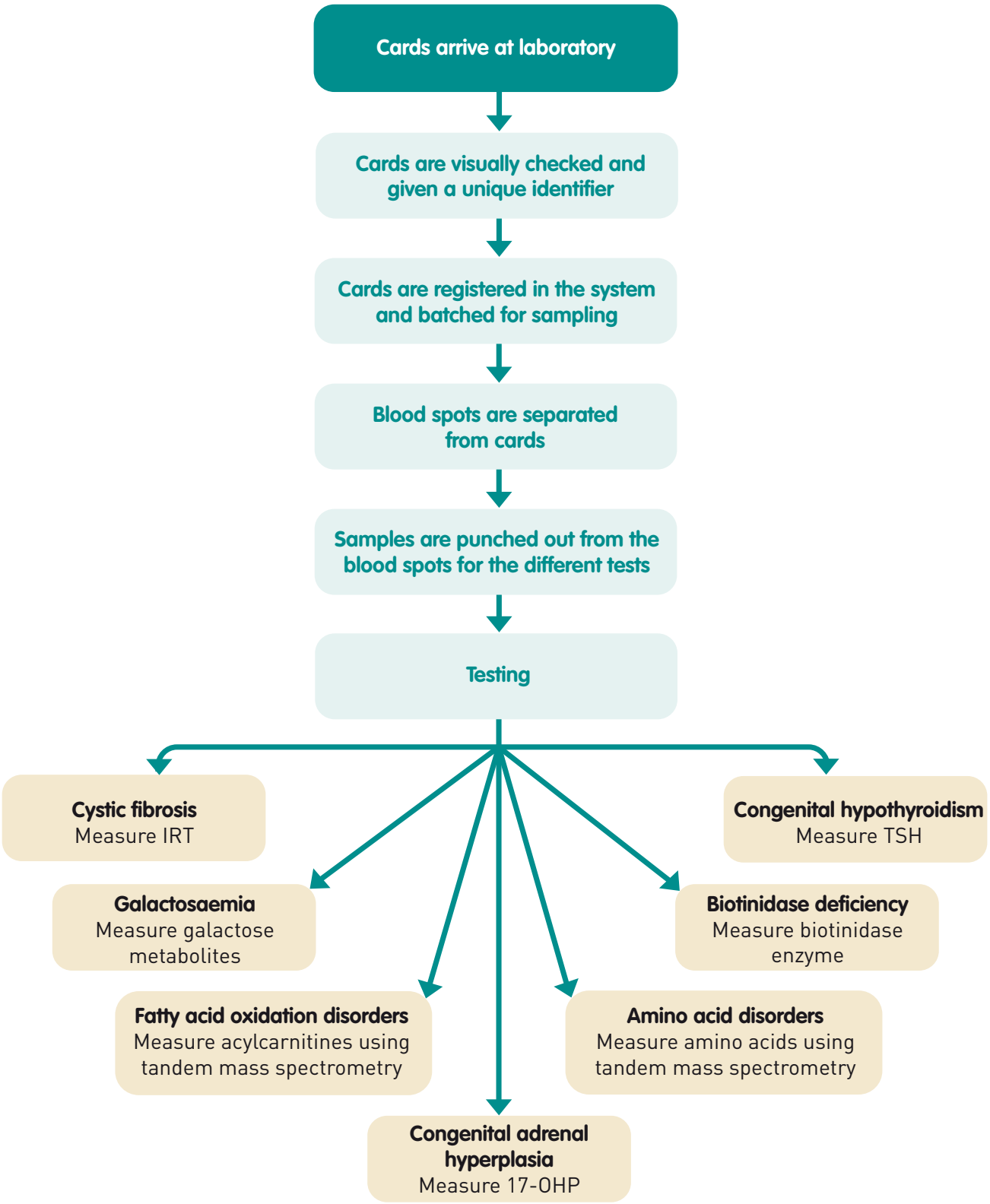
4. Send the repeat sample to the laboratory as soon as the blood is dry.

Samples must arrive at the laboratory within ten days of being requested.

5. Notify parents/guardians of the repeat sample results when they are received.

8 LABORATORY PROCESSES

When the laboratory receives blood spot cards, they are registered in the system, the blood spots are separated from the card, and testing takes place. After testing, the cards and residual blood spots are either held indefinitely in secure storage or, on request, returned to parents/guardians. Parents/guardians, or individuals, can request the return of the residual blood spots at any time.



9 SCREENING RESULTS

9.1 Results

There are three possible screening results:

- negative
- unsuitable sample
- positive.

LMCs must reconcile laboratory reports with samples they have sent for testing, and clarify any discrepancies with the laboratory.

From 2010, LMCs will receive hard copy results for every sample. If an LMC does not receive a result within 7-10 days of posting a sample, they must contact the laboratory.

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

If an LMC has clinical concerns about a baby, they must refer them to a paediatrician regardless of the screening result or whether a result has been received. If a result has not been received the LMC should also contact the laboratory.

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

9.2 Negative results

A negative screening result means that biochemical levels are within the normal range and the baby has a very low risk of having one of the disorders screened for.

A hard copy result will be sent to the LMC for every sample collected and tested. LMCs must ensure they have a result for every screened baby in their care. If a result has not been received the LMC must contact the laboratory.

9.3 Unsuitable samples

If a sample is unsuitable, this means that the sample has not been accurately tested by the laboratory, and the baby has not been fully screened for metabolic 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 send a letter requesting a repeat sample. A repeat sample must be provided so that the baby can be screened.

The LMC is responsible for taking the repeat sample, and must follow the procedure in section 7.10: Repeat sample procedure.

Unsuitable samples	
GUIDELINE	**INFORMATION**
LMCs must follow laboratory directions in the case of unsuitable samples, and take repeat samples (with informed consent) as soon as possible and ensure they reach the laboratory within ten days of the request.	Refer to section 7.9 Repeat samples.

9.4 Positive results

If a baby receives a positive screening result, LMCs must take further action, according to the advice of laboratory staff.

For CH, CAH, amino acid disorders, FAODs and galactosaemia, a positive screening result means either:

- a slightly abnormal result (testing indicates levels of certain biochemicals are slightly higher than normal; the baby is at a slightly increased risk of having a disorder) or
- a significantly abnormal result (testing indicates levels of certain biochemicals are significantly higher than normal; the baby is at a significantly increased risk of having a disorder).

For biotinidase deficiency, a positive screening result means a total deficiency of the biotinidase enzyme.

For CF, a positive screening result means a positive mutation test.

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 Metabolic Screening Programme. The laboratory can assist with further information.

Slightly abnormal results

For CH, CAH, amino acid disorders, FAODs and galactosaemia, the screening test measures the levels of different biochemicals and compares them with normal ranges expected for healthy babies.

Each disorder has a predetermined cut-off point of biochemical levels, above which a result is deemed to be outside the normal range. A result outside the normal range, but below a second, higher cut-off point, is 'slightly abnormal'. Babies with these levels are at a slightly increased risk of having a disorder.

If a result is slightly abnormal, the laboratory will send the LMC a letter requesting a repeat sample. The LMC is responsible for the process of obtaining the repeat sample, and should follow the procedure set out in section 7.10: Repeat sample procedure.

The laboratory will give guidance on the appropriate referral and level of urgency for slightly abnormal results. Babies will need diagnostic testing to confirm the presence of any disorder.

Screening for CF and biotinidase deficiency is different. See the 'Significantly abnormal results' section within this chapter for more information about screening results for these disorders.

Slightly abnormal results	
GUIDELINE	**INFORMATION**
LMCs must follow laboratory directions for slightly abnormal results, and take repeat samples (with informed consent) as soon as possible and ensure they reach the laboratory within ten days of the request.	Refer to section 7.10: Repeat sample procedure.
LMCs must communicate slightly abnormal results to parents/guardians with appropriate care, and provide information about the need for repeat samples in a way that parents/guardians can understand.	LMCs should obtain information about results from the laboratory before communicating with parents/guardians, and should clearly convey whether a repeat sample is necessary because the first sample was unsuitable, or because the screening results were abnormal. The laboratory may send information about a specific disorder in the case of slightly abnormal results. See Appendix Two: Disorders in the Newborn Metabolic Screening Programme for basic information about specific disorders, and Appendix One: Sources of support and further information for references for further information. The laboratory will assist with arranging referral to diagnostic and treatment services if this is necessary.

Significantly abnormal results

Congenital hypothyroidism, congenital adrenal hyperplasia, amino acid disorders, fatty acid oxidation disorders, and galactosaemia

A screening result for CH, CAH, amino acid disorders, FAODs or galactosaemia is 'significantly abnormal' if the biochemical levels measured are outside the higher cut-off point (that is, outside the range of 'slightly abnormal' results). Babies with these levels are at a significantly increased risk of having a disorder.

If a screening result is significantly abnormal, laboratories will advise LMCs over the telephone, and give guidance on the appropriate referral and level of urgency. Babies will need diagnostic testing to confirm the presence of any disorder.

Cystic fibrosis

Cystic fibrosis testing is a two-tier process: first, levels of immunoreactive trypsin (IRT) are measured; secondly, the 1 percent of samples with the highest IRT levels are tested for the most common CF mutations. Babies with one or two mutations produce a positive screening result, and may either have CF or be a carrier.

In the case of a positive mutation test, the laboratory will write to the LMC advising that the baby be referred to a paediatrician for diagnostic testing.

Biotinidase deficiency

Biotinidase deficiency is screened for by measuring the level of biotinidase enzyme. A total deficiency of the enzyme is a positive result. The laboratory will write to the LMC advising the result and the action the LMC should take.

Significantly abnormal results	
GUIDELINE	**INFORMATION**
LMCs must act urgently and follow laboratory directions on notifications of significantly abnormal results.	The laboratory's guidance to the LMC may include: • asking if the baby is feeding well or showing any symptoms of a disorder • providing written information about the disorder suspected • asking the LMC to examine the baby and refer to a paediatrician if they are concerned • advising that the baby be referred to a paediatrician with the screening results • advising that the baby be taken to an emergency department with the screening results. If referral to genetic services is appropriate, this will be arranged by a paediatrician.
LMCs must communicate significantly abnormal results to parents/guardians with appropriate care, and provide information about the disorder in a way that parents/guardians 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 parents/guardians.

10 DATA, INFORMATION, AND MONITORING

10.1 Data and information collection

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

BABY	SAMPLE	LMC
• 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.	• date and time of sample • laboratory-assigned identification number • whether this is the first sample • screening results • outcome of diagnostic testing • information about what has been reported and to whom • any clinical information that was advised to the laboratory with the sample • who has received reports.	• name • midwifery/medical council number or common person number • contact phone numbers • contact if LMC is going to be away.

10.2 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 any abnormal results are given to LMCs
- confirm that babies 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.

Data and information collection and use	
GUIDELINE	
LMCs must advise parents/guardians of the data and information collected about themselves and their baby, and how the NMSP may use it, including for monitoring and evaluation purposes, as part of the initial discussion during pregnancy.	

10.3 Monitoring

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

11 STORAGE, RETURN, AND FUTURE USES OF RESIDUAL BLOOD SPOTS

Parents/guardians can request that residual blood spots be returned after screening.

Please note: This section may change due to a review of the policies on storage, retention, and uses of blood spot cards. Any updates to these policies will be notified via the NSU's website www.nsu.govt.nz

11.1 Storage or return of residual blood spots

When screening is complete, if a parent/guardian has not requested that the residual blood spots be returned, they are held indefinitely in secure storage.

Parents/guardians should understand the options for storage or return of residual blood spots as part of the offer of screening (see section 6.5: Offer of screening). LMCs must discuss with parents/guardians whether they want to accept or deline 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 parents/guardians, 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.

Figure 6: Residual blood spots after laboratory testing

LMCs should advise parents/guardians that they can request the return of their baby's 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 section 11.2 Uses of residual blood spots after screening.

Return of residual blood spots	
GUIDELINE	**INFORMATION**
LMCs must provide parents/guardians with sufficient information to request the return of their baby's 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. **Requests for return after sample sent to laboratory** Parents/guardians should make a request for the return in writing using the 'Return of Newborn Metabolic Screening Samples (Guthrie Cards) to Family' form, available on the NSU website at www.nsu.govt.nz/Files/ANNB/guthrie_card_return_july_09.pdf Details of who may request the residual blood spots are on the form. Photographic identification and other documentation is required to ensure the laboratory sends the card to the person authorised to receive it. The laboratory must return requested blood spots to parents/guardians or individuals by tracked courier within one month of the request.

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

11.2 Uses of residual blood spots after screening

Residual blood spots may be used to improve programme quality, for example through investigation of false positive and false negative results, quality assurance processes, and improving programme equipment and testing.

Residual blood spots may also be used for:

- investigation of illness or deaths related to specific genetic conditions or infections
- research approved by an ethics committee towards improving the health of babies and their families
- assisting police to identify victims (of accidents, crimes or natural disasters)
- other testing, with authorised consent
- testing ordered by a court.

11.3 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. Parents/guardians should be advised of this during the initial discussion (see section 6.4: Initial discussion).

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

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

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

To date, research using residual blood spots has not been undertaken. However, there is potential for this to happen in the future. This may occur as part of a research programme approved by an ethics committee.

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

The Ministry of Health has a Memorandum of Understanding (MoU) with the New Zealand Police regarding the use of blood spots in exceptional circumstances. These circumstances have included identifying victims of natural disasters (eg Tsunamis and house fires). The Memorandum can be viewed on the NSU's website at www.nsu.govt.nz

Possible future uses of residual blood spots	
GUIDELINE	**INFORMATION**
LMCs must advise parents/guardians about the possible future uses of their baby's residual blood spots if they are stored.	Parents/guardians must be advised that stored residual blood spots may be used in the future for the purposes set out in this section.

Appendices

APPENDIX ONE: SOURCES OF SUPPORT AND FURTHER INFORMATION

LabPLUS
Auckland City Hospital
Auckland District Health Board
PO Box 872
Auckland
Phone: 09 307 4949 extn 6759
Toll free: 0800 522 7587
Email: ntc@adhb.govt.nz
http://www.labplus.co.nz

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

National Screening Unit
Newborn Metabolic Screening Programme
Private Bag 92522
Wellesley Street
Auckland
Phone: 09 580 9000
Fax: 09 580 9001
http://www.nsu.govt.nz

Kathy Bendikson, Programme Manager
kathy_bendikson@moh.govt.nz

National Screening Unit Resources
Available from: http://www.nsu.govt.nz

Resources for health practitioners

- Best Practice for Health Practitioners (DVD).
- Best Practice Key Messages.
- Blood Sample Protocol (Heel Prick) for Level 2 Special Care Baby Units (SCBU).
- Blood Sample Protocol (Heel Prick) for Level 3 Neonatal Intensive Care Units (NICU).
- Blood spot cards – the impact of correct sample taking.
- Heel pricks – warming, pain relief and lancet use.

Fact sheets for parents/guardians

- Congenital hypothyroidism.
- Congenital adrenal hyperplasia.
- Medium chain acyl-CoA dehydrogenase (MCAD) deficiency.

Resources available from the laboratory for confirmed cases

- Phenylketonuria handbook.
- Fatty acid oxidation disorders handbook.

New Zealand Organisation for Rare Disorders (NZORD)

PO Box 38-538
Petone 5045
Phone: 04 471 2226
Email: enquiries@nzord.org.nz
http://www.nzord.org.nz

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

Genetic Services, New Zealand

Northern and Midland Region
Toll free: 0800 476 123

Central Region
Toll free: 0508 364 436

Southern Region
Toll free: 0508 364 436

Health and Disability Commissioner

http://www.hdc.org.nz

Language Line

Language Line is a telephone interpreting service, available from Monday to Friday 9am - 6pm through participating agencies. For a list of participating agencies and the 40 languages available, go to
http://www.dia.govt.nz/oeawebsite.nsf/wpg_URL/language-line-Index
or contact
Email: languageline@dia.govt.nz

Office of the Privacy Commissioner

http://www.privacy.org.nz

Ministry of Health

Primary Maternity Services Notice 2007:
http://www.moh.govt.nz/moh.nsf/indexmh/maternity-section88notice

Refugee Council

http://www.rc.org.nz

Tamariki Ora

http://www.wellchild.org.nz/index.htm

Other sources/resources

New South Wales Centre for Genetic Education
www.genetics.edu.au

Journal of the American Academy of Pediatrics
www.pediatrics.org

Genetics and metabolic disease articles
http://emedicine.medscape.com/pediatrics_genetics

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

Screening: Evidence and practice
Raffle A, Gray JAM. 2007. Oxford: Oxford University Press

United States National Center for Biotechnology Information's 'GeneReviews' database
(provides 'expert-authored, peer-reviewed, current disease descriptions that apply genetic testing to the diagnosis, management, and genetic counselling of patients and families with specific inherited conditions')
http://www.ncbi.nlm.nih.gov/sites/GeneTests/review?db=GeneTests

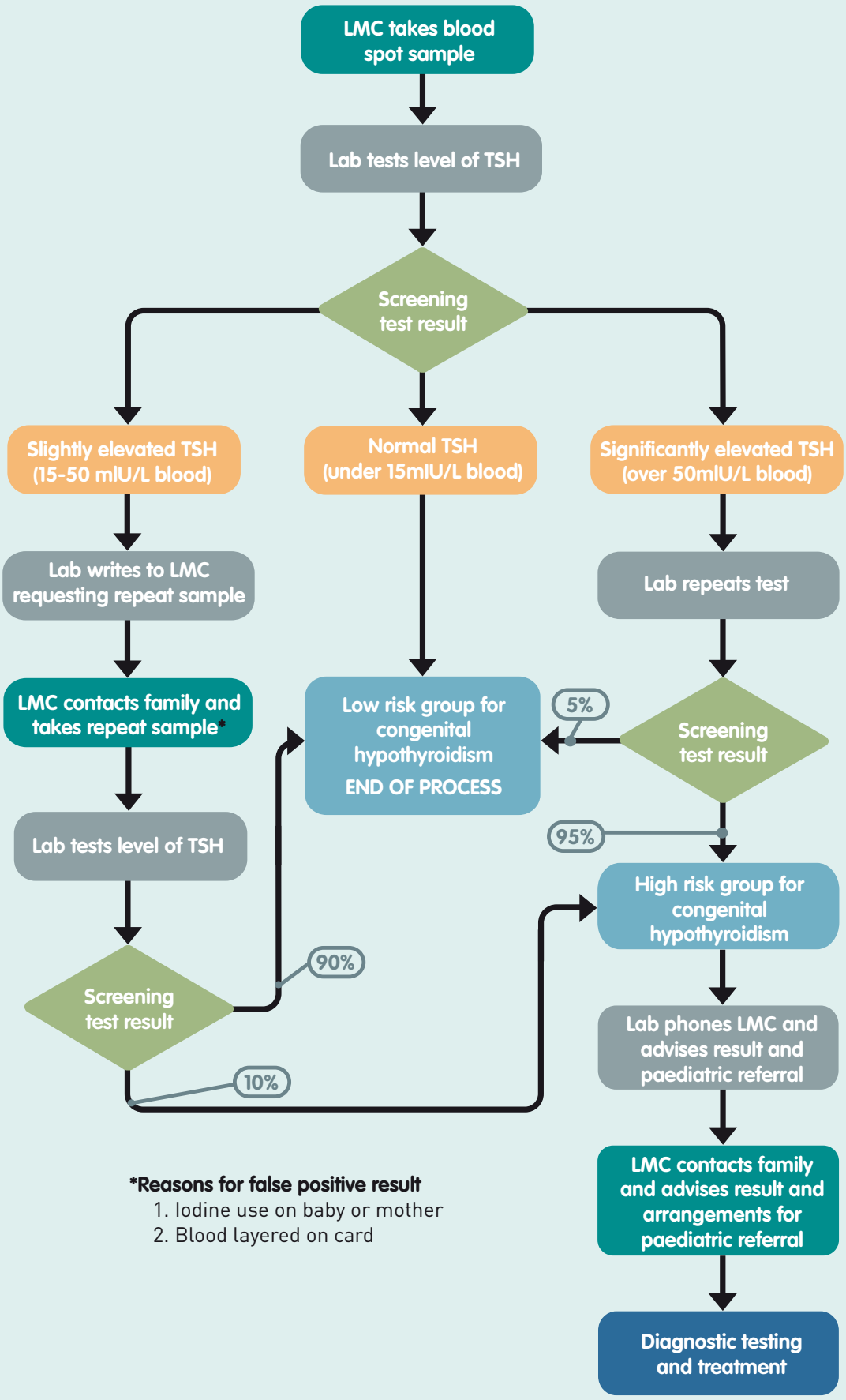
United States National Library of Medicine's Genetics Home Reference service
(provides 'consumer-friendly information about the effects of genetic variations on human health')
http://www.ghr.nlm.nih.gov/

APPENDIX TWO: DISORDERS IN THE NEWBORN METABOLIC SCREENING PROGRAMME

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	1/4000: approx 15 babies every year
Cystic fibrosis	A defective gene and its protein product lead to thick sticky mucus.	Medication and physiotherapy	1/7000: approx 8 babies every year
Amino acid disorders (14 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 developmental delay).	Special diet	1/12,000: approx 5 babies every year
Fatty acid oxidation disorders (FAODs) (9 disorders), 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)	1/12,000: approx 5 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 salt/hormonal imbalances in both sexes.	Hormone replacement	1/20,000: 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	1/100,000: 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)	1/150,000: approx 1 baby every 3 years

*For the most up-to-date incidences see www.nsu.govt.nz

Congenital hypothyroidism screening



***Reasons for false positive result**

1. Iodine use on baby or mother
2. Blood layered on card

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.

A baby with congenital hypothyroidism may have no obvious symptoms. However, some babies 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 babies correctly treated with thyroxine can grow and develop normally.

Screening

Blood is tested for levels of thyroid stimulating hormone (TSH). Babies 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.

Action

Slightly abnormal result

If LMCs receive a letter from the laboratory advising of a slightly abnormal TSH result, they should:

- contact the laboratory if they require further information
- contact the family and inform them of the screening result
- evaluate the baby: although they may display no obvious symptoms, they may be very sleepy and feed slowly, be constipated, or have prolonged jaundice
- with family consent, take a repeat sample and send it to the laboratory as soon as possible
- support the family by providing information about the reasons for a slightly abnormal result, and the disorder
- notify the family of the results of the repeat sample.

Significantly abnormal result

If LMCs are phoned by the laboratory and informed of a significantly abnormal TSH result, they should:

- follow any advice given by the laboratory
- arrange a paediatric referral (if not organised by the laboratory)
- contact the family immediately and inform them of the screening result and the arrangements for paediatric assessment
- support the family by providing information about the reasons for a significantly abnormal result, diagnostic testing and the disorder.

Treatment

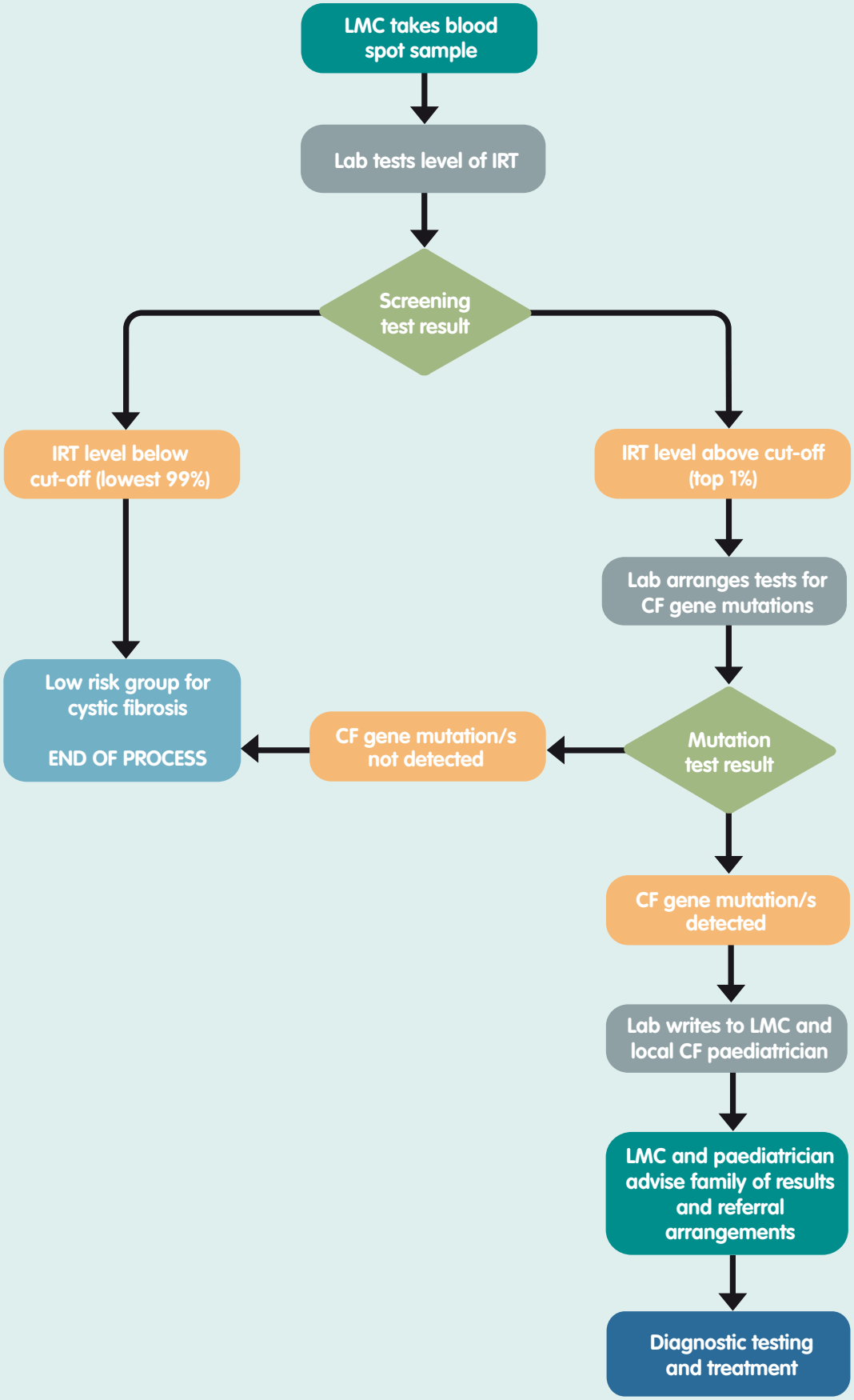
Babies with CH are cared for by a paediatrician (usually an endocrinologist). Treatment involves a daily oral dose of thyroxine for life (this is given to babies as a suspension), in sufficient quantities to bring thyroxine levels up to normal. Thyroxine levels are monitored with regular blood tests, 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.

Maternal hypothyroidism

There is a small possibility that abnormalities in babies may be masked by the mother's condition. Full thyroid function tests at one week of age is recommended for all babies with a mother with hypothyroidism.

Cystic fibrosis screening



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 parents/guardians and siblings to be aware of their potential carrier status and make informed choices about their families.

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 New Zealand. There are more than 700 possible CF mutations.

Babies in whom one or two mutations are present produce a positive screening result. The notification letter to the LMC will report the presence of one or two mutations, and advise that the baby may either have CF or be a carrier and that further testing will determine this.

LMCs should refer babies who test positive for CF to a paediatrician for full assessment and diagnostic testing as soon as possible. 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, tests for faecal chymotrypsin).

Action

Positive mutation screening test

If LMCs receive a letter from the laboratory advising of a positive mutation screening test result, they should:

- contact the laboratory if they require further information
- contact the family and inform them of the screening result and that further testing is required to confirm the CF status – the screening result is not a diagnostic confirmation of CF
- provide the family with the information on CF sent with the notification letter
- discuss diagnostic testing with the family, as in the information sent with the notification letter
- contact a local CF paediatrician
- support the family by providing further information if required.

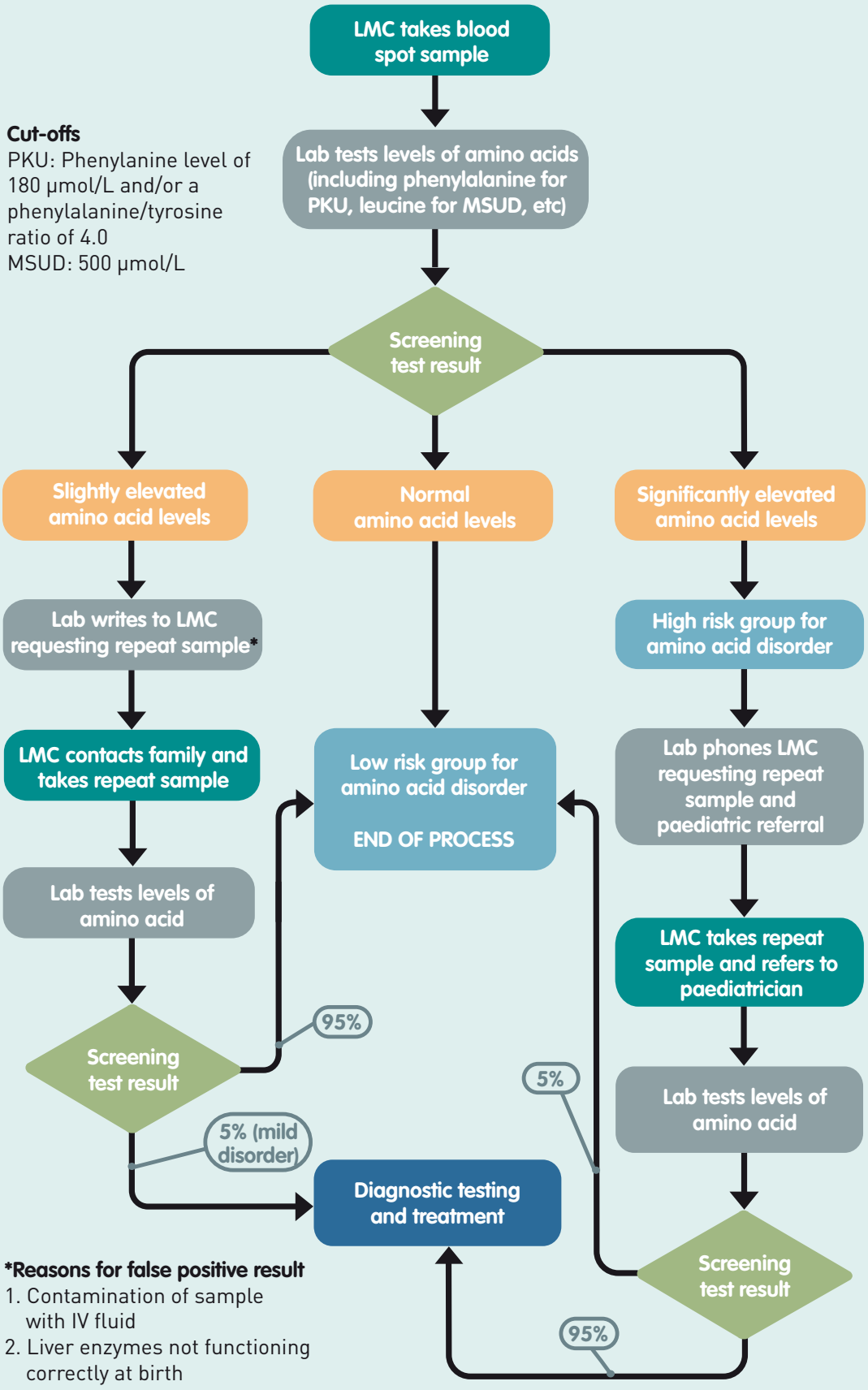
Treatment

Babies 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. 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.

Family history of cystic fibrosis

If there is a family history of CF, consultation with a CF paediatrician is recommended. A negative newborn screening result does not give information about CF carrier status as the sample may not have had mutation testing.

Amino acid disorder screening



Cut-offs
PKU: Phenylanine level of 180 µmol/L and/or a phenylalanine/tyrosine ratio of 4.0
MSUD: 500 µmol/L

***Reasons for false positive result**
1. Contamination of sample with IV fluid
2. Liver enzymes not functioning correctly at birth
3. Liver disease

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 building muscle, 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 14 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 hydroxlyase. This enzyme is needed to process the amino acid phenylalanine (most commonly found in protein-rich foods such as dairy products, red meat, chicken, fish, eggs, nuts, beans and lentils).

The phenylalanine which can not be metabolised builds up in blood and tissues. This leads to toxicity, and can prevent the brain from developing normally. Lack of phenylalanine hydroxlyase 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, babies with PKU can grow and develop to their full potential.

Screening

Blood is tested for levels of phenylalanine and tyrosine.

Action

Slightly abnormal result

If LMCs receive a letter from the laboratory advising of a slightly abnormal PKU result, they should:

- contact the laboratory if they require further information
- contact the family and inform them of the screening result
- evaluate the baby: a baby with PKU may not have any obvious symptoms, as the excess phenylalanine will not have yet built up sufficiently to cause damage
- with family consent, take a repeat sample and send it to the laboratory as soon as possible
- support the family by providing information about the reasons for a slightly abnormal result and the disorder
- notify the family of the results of the repeat sample.

Significantly abnormal result

If LMCs are phoned by the laboratory and informed of a significantly abnormal PKU result, they should:

- arrange an urgent paediatric referral
- contact the family immediately and inform them of the screening result and arrangements for paediatric assessment
- with family consent, take a repeat sample and send it to the laboratory as soon as possible
- support the family by providing information about the reasons for a significantly abnormal result, diagnostic testing and the disorder.

Treatment

Babies 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.

The nutritional supplement contains all the essential nutrients the baby needs, including amino acids, except phenylalanine, and some breast milk. Phenylalanine tolerance among babies 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 form of MSUD, low muscle tone, lethargy, poor feeding and hypoglycaemia develop in the first week. Without treatment, the baby 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, babies with MSUD can grow and develop to their full potential.

Screening

Blood is tested for levels of leucine, isoleucine, and valine.

Action

Slightly abnormal result

If LMCs receive a letter from the laboratory advising of a slightly abnormal MSUD result, they should:

- contact the laboratory if they require further information
- contact the family and inform them of the screening result
- evaluate the baby: babies with MSUD will not show any symptoms at first, but low muscle tone, lethargy, poor feeding and low blood sugar levels may develop in the first week of life
- with family consent, take a repeat sample and send it to the laboratory as soon as possible
- support the family by providing information about the reasons for a slightly abnormal result and the disorder
- notify the family of the results of the repeat sample.

Significantly abnormal result

If LMCs are phoned by the laboratory and informed of a significantly abnormal MSUD result, they should:

- be guided by the laboratory staff regarding the baby's condition
- contact the family immediately to inform them of the screening result and any arrangements discussed with the laboratory
- with family consent, take a repeat sample and send it to the laboratory as soon as possible
- support the family by providing information about the reasons for a significantly abnormal result, diagnostic testing and the disorder.

Treatment

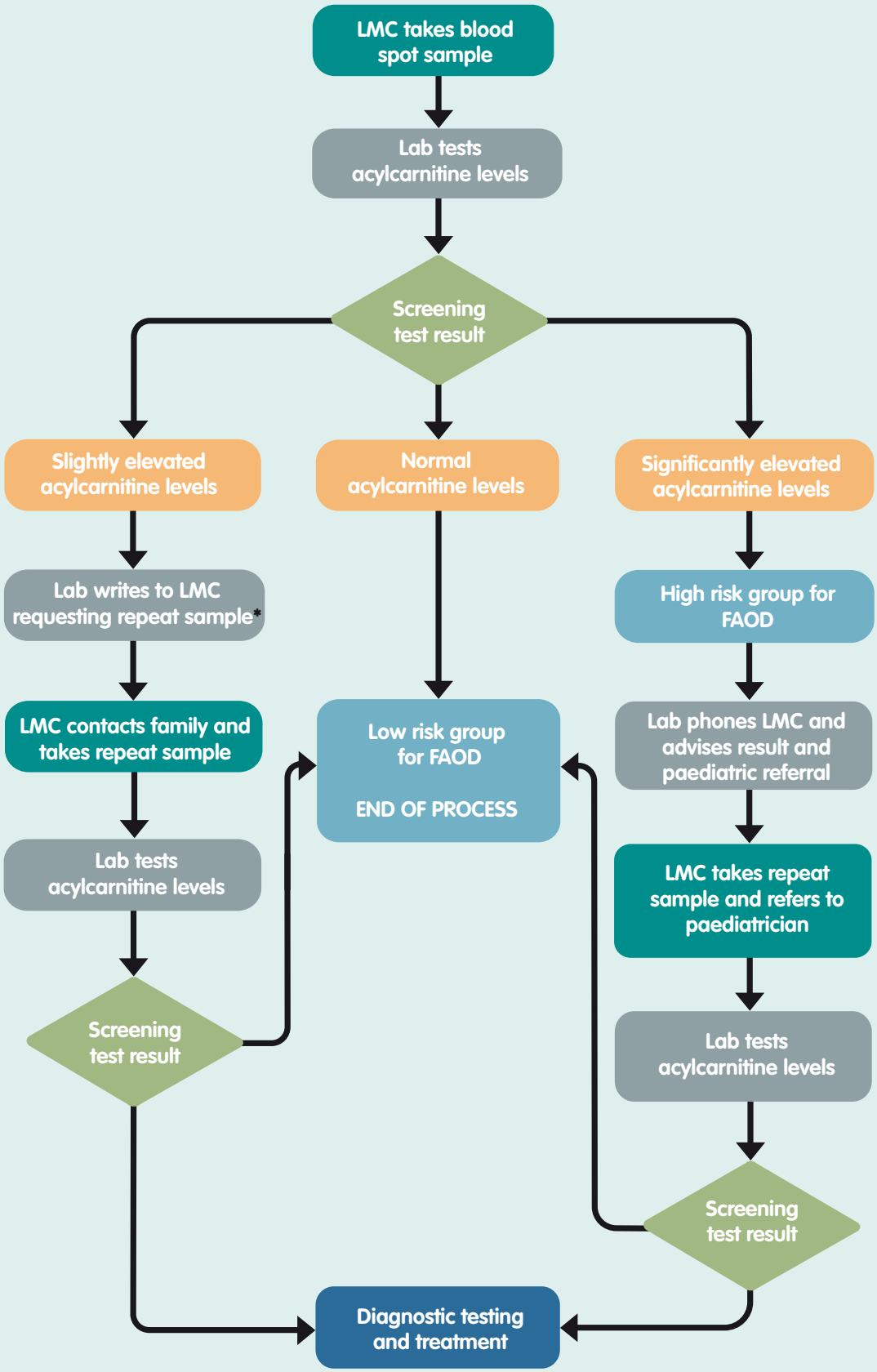
Babies with MSUD are cared for by a clinical team which includes a metabolic paediatrician, dietician, nurse and local paediatrician if necessary. MSUD is treated with a low-protein diet and a special nutritional supplement, both of which need careful monitoring. Regular blood tests to measure branched-chain 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, citrin deficiency).
- Glutaric acidaemia type I (glutaryl-CoA dehydrogenase deficiency).
- Homocystinuria (cystathionine beta-synthase deficiency).
- 3-hydroxy-3-methylglutaryl-CoA lyase deficiency (HMGCoA lyase deficiency).
- Isovaleric acidaemia (isovaleryl-CoA dehydrogenase deficiency).
- Beta-ketothiolase deficiency (mitochondrial acetoacetyl-CoA thiolase deficiency).
- 3-methylcrotonyl-CoA carboxylase deficiency.
- Methylmalonic acidurias (mutase deficiency, CblA, CblB, CblC, CblD defects).
- Multiple carboxylase deficiency (holocarboxylase synthetase deficiency).
- Propionic acidaemia (propionyl-CoA carboxylase deficiency).
- Tyrosinaemia (fumaryl acetoacetase deficiency, tyrosine aminotransferase deficiency).

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

Fatty acid oxidation disorder screening



***Reasons for false positive result**
Too much blood on the card

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 can not 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 families to ensure that babies do not fast too long and have treatment regimes in place if babies 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).

Action

Slightly abnormal result

If LMCs receive a letter from the laboratory advising of a slightly abnormal FAOD result, they should:

- contact the laboratory if they require further information
- contact the family and inform them of the screening result
- evaluate the baby: a baby with MCAD deficiency may not show any symptoms unless they are not feeding well (for example, due to another illness) and the dietary and liver sources of energy are exhausted, leading to low blood glucose levels
- with family consent, take a repeat sample and send it to the laboratory as soon as possible
- support the family by providing information about the reasons for a slightly abnormal result and the disorder
- notify the family of the results of the repeat sample.

Significantly abnormal result

If LMCs are phoned by the laboratory and informed of a significantly abnormal FAOD result, they should:

- be guided by the laboratory staff regarding the baby's condition
- contact the family immediately and inform them of the screening result and any arrangements as discussed with the laboratory
- with family consent, take a repeat sample and send it to the laboratory as soon as possible
- support the family by providing information about the reasons for a significantly abnormal result, diagnostic testing and details of the disorder.

Treatment

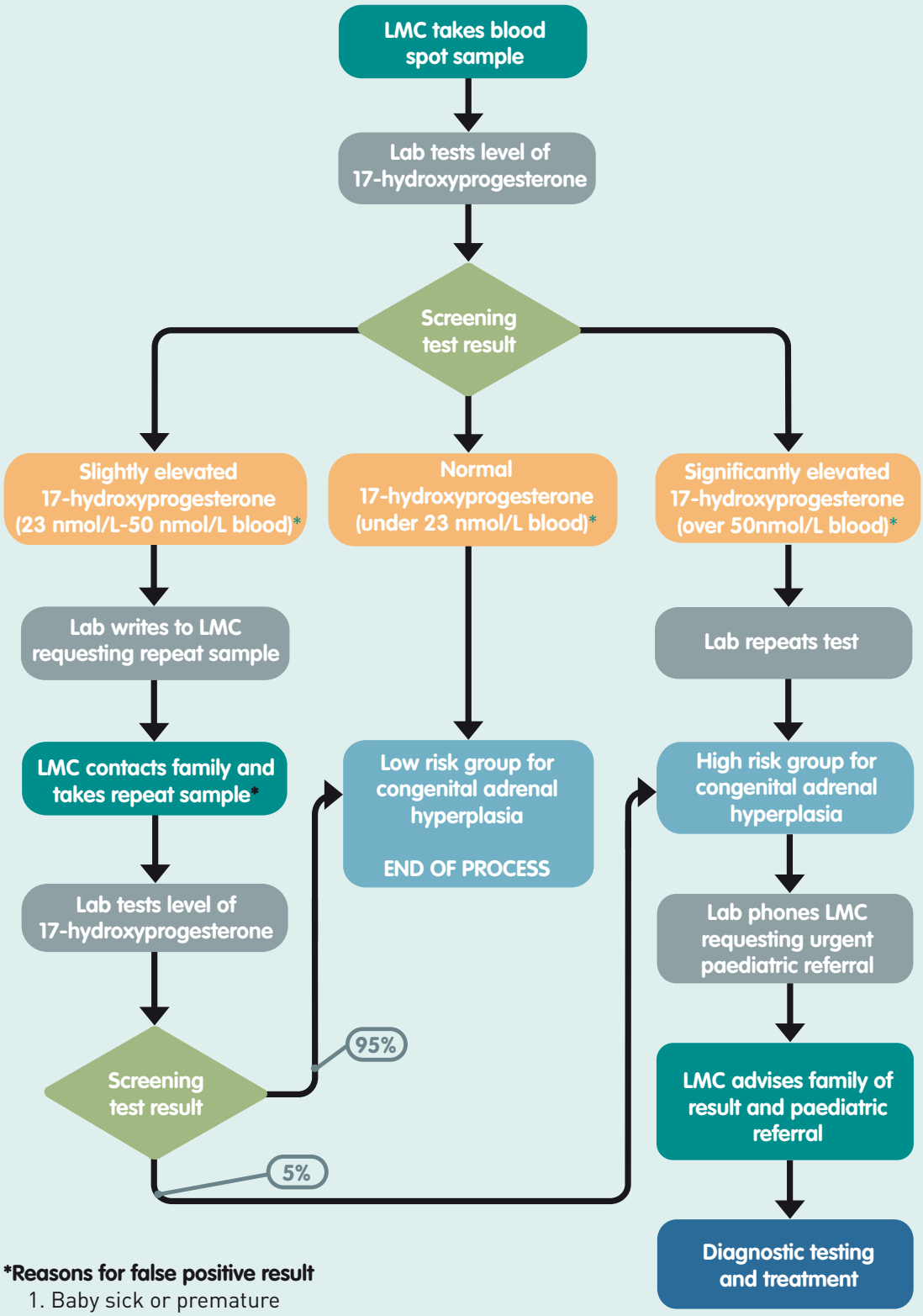
Babies with FAODs are cared for by a metabolic paediatrician. They will need to attend a metabolic clinic and take vitamin supplements. Treatment may be as simple as family advice about regular feeding and identification of symptoms. In the case of concurrent illnesses or vomiting, a care plan will include 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 transporter defect.
- Carnitine palmitoyltransferase-I deficiency (CPT-I).
- Carnitine palmitoyltransferase-II deficiency (CPT-II).
- 3-hydroxy long-chain acyl-CoA dehydrogenase deficiency (LCHAD).
- Trifunctional protein deficiency (TFP).
- Multiple acyl-CoA dehydrogenase deficiency (MADD).
- Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD).

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

Congenital adrenal hyperplasia screening



***Reasons for false positive result**

1. Baby sick or premature
2. Blood layered on card

*Cut-off levels are different for low birth weight babies

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

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

Babies 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).

Action

Slightly abnormal result

If LMCs receive a letter from the laboratory advising of a slightly abnormal CAH result, they should:

- contact the laboratory if they require further information
- contact the family and inform them of the screening result
- evaluate the baby: symptoms may include poor feeding, vomiting, diarrhoea, failure to thrive, lethargy, dehydration and weak cry
- with family consent, take a repeat sample and send it to the laboratory as soon as possible
- support the family by providing information about the reasons for a slightly abnormal result and the disorder
- notify the family of the results of the repeat sample.

Significantly abnormal result

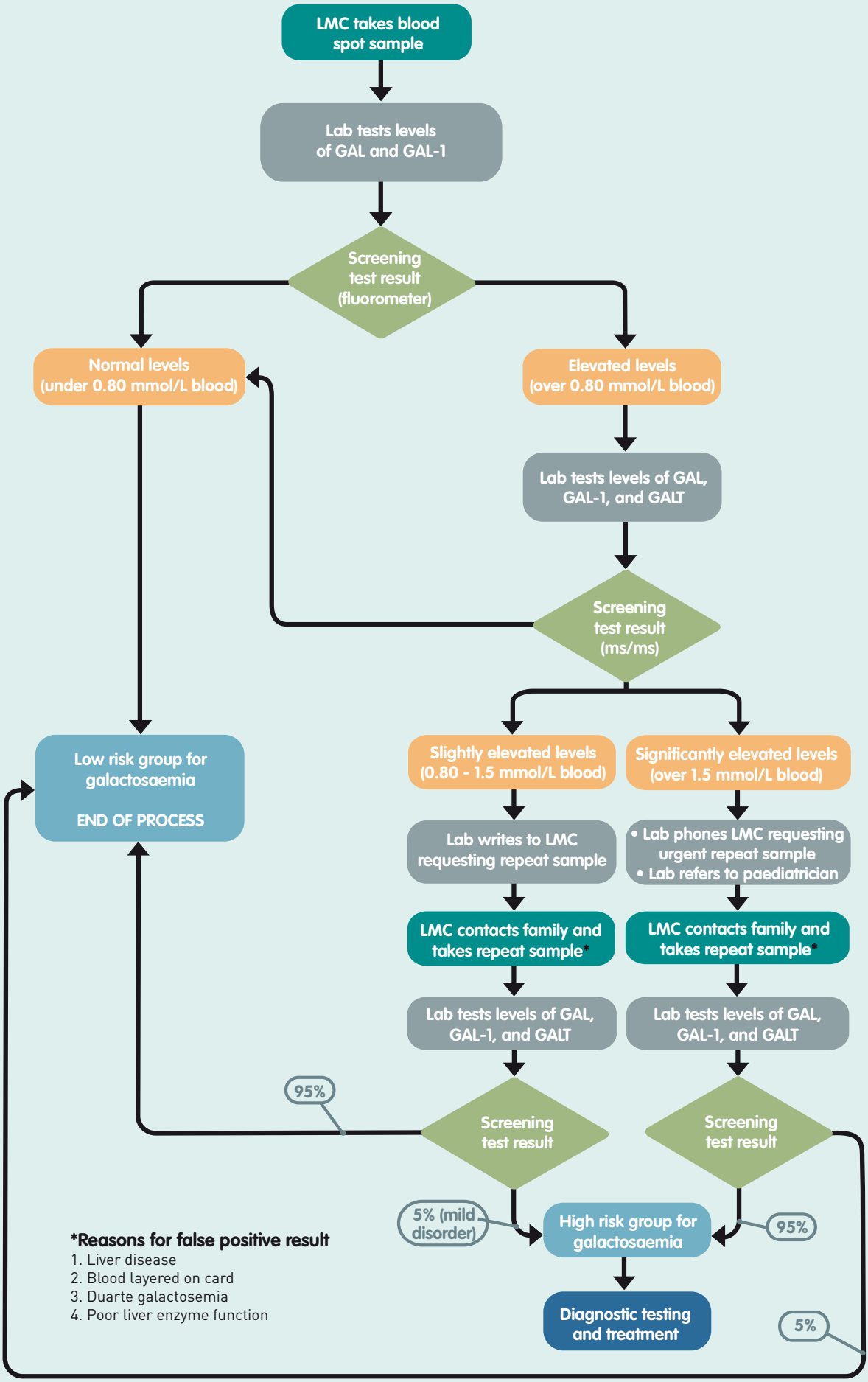
If LMCs are phoned by the laboratory and informed of a significantly abnormal CAH result, they should:

- arrange an urgent paediatric referral
- contact the family immediately and inform them of the screening result and the arrangements for paediatric assessment
- support the family by providing information about the reasons for a significantly abnormal result, diagnostic testing and the disorder.

Treatment

Babies 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 androgen, which occur in the presence of reduced cortisol levels.

Galactosaemia screening



***Reasons for false positive result**

1. Liver disease
2. Blood layered on card
3. Duarte galactosemia
4. Poor liver enzyme function

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. Babies 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, babies 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.

One mild variant of the galactosaemia gene, the Duarte gene, is common. Among carriers of one Duarte and one normal gene, enzyme activity happens at about 75 percent of normal levels. Among compound heterozygotes (carriers of one galactosaemia gene and one Duarte gene) enzyme activity happens at about 25 percent of normal levels. Incidence of this is about one in five hundred. In some cases, a screening test may indicate abnormal galactose metabolites in a baby's blood and reduced levels of enzyme activity, but the baby will not display any symptoms, be lactose intolerant 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 in the blood (galactose and galactose-1-phosphate).

Action

Slightly abnormal result

If LMCs receive a letter from the laboratory advising of a slightly abnormal galactosaemia result, they should:

- contact the laboratory if they require further information
- contact the family and inform them of the screening result
- evaluate the baby: some babies may show no symptoms, but if the disorder is more severe, symptoms may include vomiting, diarrhoea, failure to thrive, hepatomegaly, ascites and cataracts
- with family consent, take a repeat sample and send it to the laboratory as soon as possible
- support the family by providing information about the reasons for a slightly abnormal result and the disorder
- notify the family of the results of the repeat sample.

Significantly abnormal result

If LMCs are phoned by the laboratory and informed of a significantly abnormal galactosaemia result, they should:

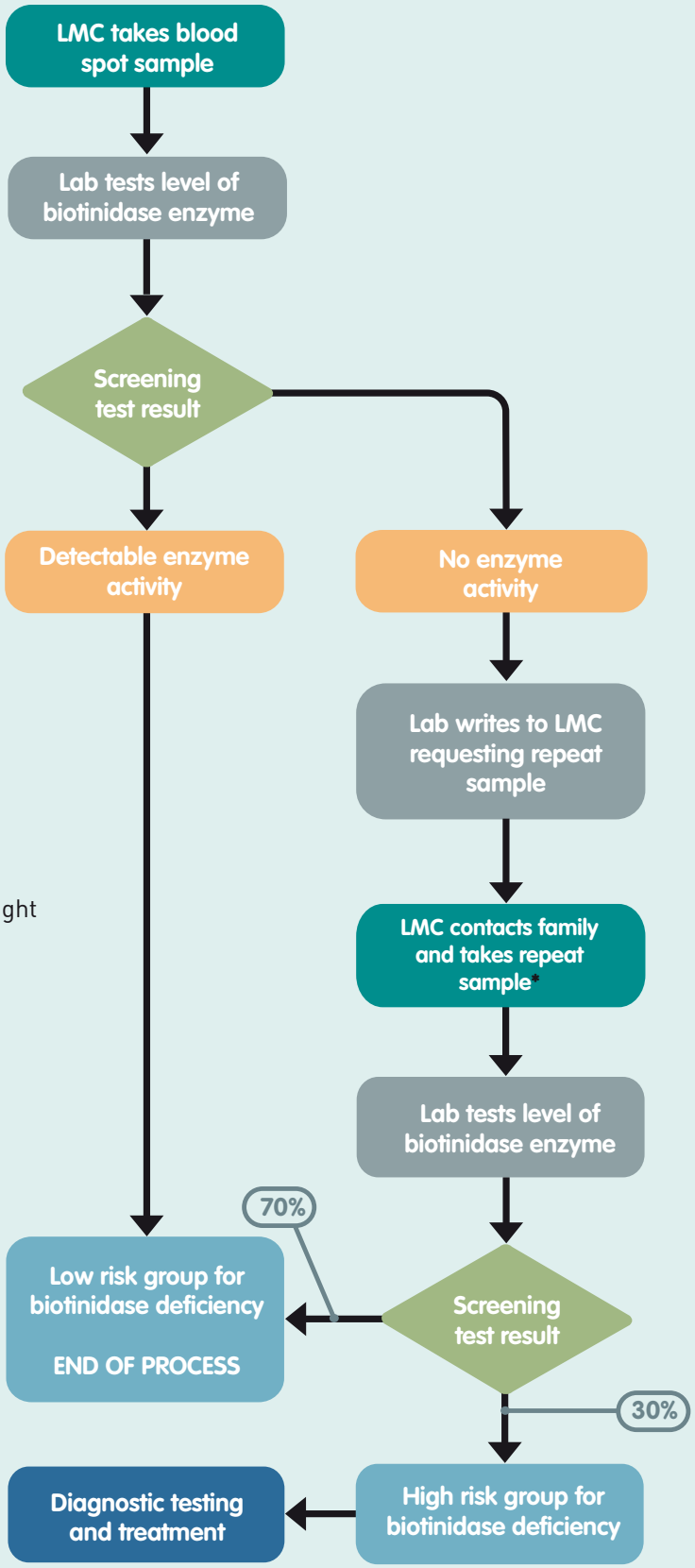
- be guided by the laboratory staff regarding the baby's condition: if a baby is symptomatic they may advise immediate paediatric referral
- contact the family immediately and inform them of the screening result
- with family consent, take a repeat sample and send it to the laboratory as soon as possible
- support the family by providing information about the reasons for a significantly abnormal result, diagnostic testing and the disorder.

Treatment

Babies 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. The baby 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



***Reasons for false positive result**
Sample damaged by heat or sunlight

Biotinidase deficiency screening

Description

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

Biotin is obtained from the diet, or by reuse of 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.

Babies are born with biotin stores and carboxylase enzymes, and therefore symptoms do not usually appear until the baby 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 babies correctly treated with biotin can grow and develop normally.

Screening

Blood is tested for biotinidase enzyme.

Action

Abnormal result

If LMCs receive a letter from the laboratory advising of an abnormal biotinidase result, they should:

- contact the laboratory if they require further information
- contact the family and inform them of the screening result
- evaluate the baby: symptoms will not generally appear until the baby has used up all their carboxylase enzymes, usually several months after birth
- with family consent, take a repeat sample and send it to the laboratory as soon as possible
- support the family by providing information about the reasons for an abnormal result and the disorder
- notify the family of the results of the repeat sample.

Treatment

Babies 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 affects.

APPENDIX THREE: BLOOD SPOT CARDS


NTC USE ONLY
DO NOT COVER
NHI #
BABY SURNAME
NTC USE ONLY
DO NOT COVER
BABY INFORMATION - FILL OUT OR USE LABEL
NHI Number
Surname
First Name
Sex Ethnicity:
MOTHER'S INFORMATION - if not on label
Surname
First Name
LMC INFORMATION
Name Regist. No.
Address
Mobile, Pager or Phone No.
Where was baby born
Birth time Birth date
Gestational age wk Birth wt. g
Collection time Collection date
Baby 48 hours old (minimum) Y N (circle correct answer) 1st sample Y N

This is the standard buff/beige blood spot card used for babies who are over 1500g birthweight and are not in a Neonatal Intensive Care Unit (NICU) on the day the sample is taken.


BABY NAME
NTC USE ONLY
NHI #
NTC USE ONLY
BABY INFORMATION - USE LABEL
NHI Number
Surname
First Name
Sex
MOTHER'S INFORMATION - if not on label
Surname First Name
REPORT TO
Name or role
COPY TO (LMC OR WELL CHILD PROVIDER)
Name Address
Hospital Ethnicity
Birth Time: Birth Date:
Birth Wt. (g) Gestation age wk
Collection Time: Collection Date:
Collect 1st 2nd 3rd sample

This is the blue card used for babies who are inpatients in a NICU or Special Care Baby Unit (SCBU) at the time of screening, or who are under 1500g birthweight.

APPENDIX FOUR: SCBU AND NICU SCREENING PROTOCOLS

Newborn Metabolic Screening Programme (NMSP) Blood sample protocol (heel prick) for level 2 Special Care Baby Units (SCBU)

Scope of the protocol

This protocol describes screening for specific metabolic disorders in newborns that are admitted to SCBU.

Overview of the screening protocol

Metabolic screening protocol in New Zealand to date is to obtain a heel prick sample from all babies at 48 hours when consent has been obtained from the parents/guardians.

Many very low birth weight and sick babies have false positive screens for congenital adrenal hyperplasia due to their immaturity at 48 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 abnormal level of TSH in response to a low thyroxine level. Therefore, a revised protocol for SCBU babies has been developed in consultation with NZ endocrinologists, paediatricians and neonatologists.

Due to updated screening technology it is noted that a specific amount of feeding is no longer required.

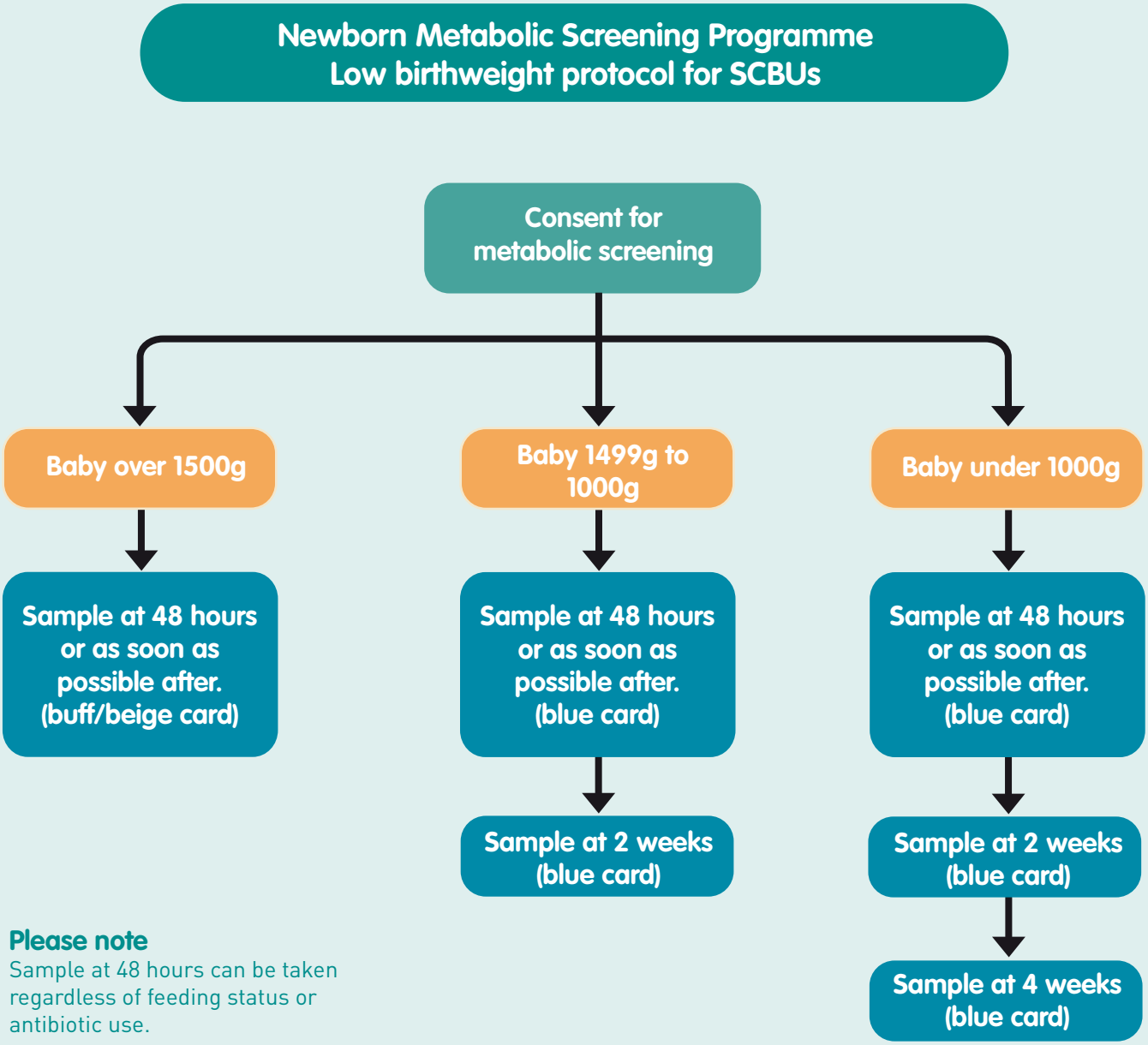
General principles

- Blue coloured blood spot cards are to be used for babies in level 2 SCBU when the babies are less than 1500g birthweight or when the screening result would have high clinical utility. Use buff/beige cards for the other babies
- A sample from all babies at 48 hours of age or as soon as possible thereafter
- A second sample at 2 weeks for babies with a birthweight less than 1500g
- A third sample at 4 weeks for babies with a birthweight less than 1000g
- Results highly suggestive of a screened disorder will be phoned and slightly abnormal results will be advised in writing
- Reporting will be to the individual(s) and/or role named on the card

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 www.nsu.govt.nz

Any queries regarding this protocol, please call Dr Dianne Webster at the National Testing Centre on 09 307 4949 x 23019.



Please note
Sample at 48 hours can be taken regardless of feeding status or antibiotic use.

Newborn Metabolic Screening Programme (NMSP)
Blood sample protocol (heel prick) for level 3 Neonatal Intensive Care Units (NICU)

Scope of the protocol

This protocol describes screening for specific metabolic disorders in newborns that are admitted to NICU.

Overview of the screening protocol

Metabolic screening protocol in New Zealand to date is to obtain a heel prick sample from all babies at 48 hours when consent has been obtained from the parents/ guardians.

Many very low birthweight and sick babies have false positive screens for congenital adrenal hyperplasia due to their immaturity at 48 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 abnormal level of TSH in response to a low thyroxine level. Therefore, a revised protocol for NICU babies has been developed in consultation with NZ endocrinologists, paediatricians and neonatologists.

Due to updated screening technology it is noted that a specific amount of feeding is no longer required.

General principles

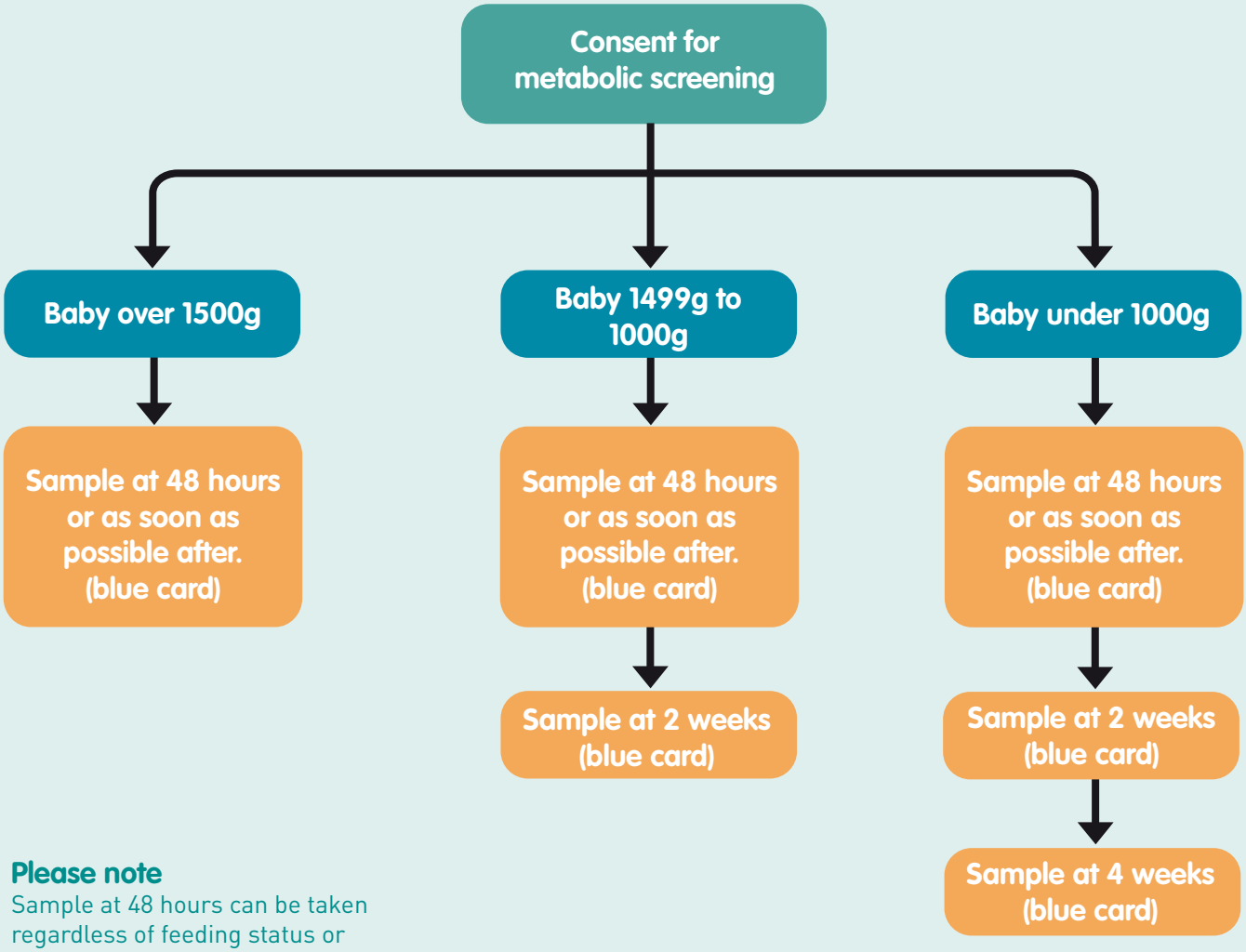
- Blue coloured blood spot cards are to be used for all babies in level 3 NICU
- A sample from all babies at 48 hours of age or as soon as possible thereafter
- A second sample at 2 weeks for babies with a birthweight less than 1500g
- A third sample at 4 weeks for babies with a birthweight less than 1000g
- Results highly suggestive of a screened disorder will be phoned and slightly abnormal results will be advised in writing
- Reporting will be to the individual(s) and/or role named on the card

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 www.nsu.govt.nz

Any queries regarding this protocol, please call Dr Dianne Webster at the National Testing Centre on 09 307 4949 x 23019.

Newborn Metabolic Screening Programme
Low birthweight protocol for NICUs



Please note
Sample at 48 hours can be taken regardless of feeding status or antibiotic use.
Please use the blue blood spot cards for all samples.

Newborn Metabolic Screening Programme (NMSP) Blood sample protocol (heel prick) for babies who have received blood transfusions

Scope of the protocol

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

Overview of the screening protocol

Metabolic screening protocol in New Zealand to date is to obtain a heel prick sample from all babies at 48 hours when consent has been obtained from the parents/ guardians. 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 48 hours of age regardless of whether or not 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 www.nsu.govt.nz.

Any queries regarding this protocol, please call Dr Dianne Webster at the National Testing Centre on 09 307 4949 x 23019.


MINISTRY OF
HEALTH
MANATŪ HAUORA