

Your Newborn Baby's Blood Test

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Outline



- Background – newborn screening in New Zealand – why develop a new information sheet?
- Development process
- The new brochure



- 4.4 M population
- 65000 births per year
 - European 54%
 - Maori 22%
 - Pacific peoples 11%
 - Asian 10%
 - Other 3%
- All screening, diagnosis and treatment government funded

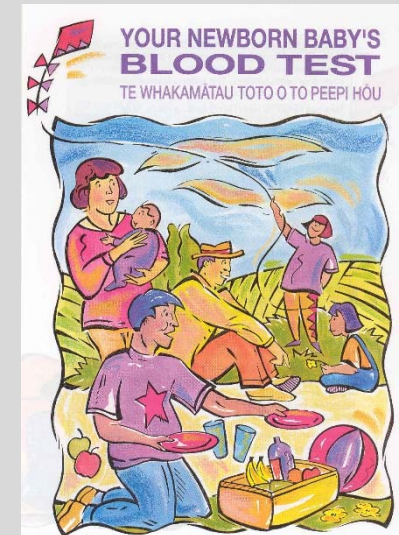
- National Screening Unit
- Specialist unit within Ministry of Health
- Funds screening through contract with local hospital laboratory



The screenshot displays the National Screening Unit website. The main navigation menu includes: Home, About Us, Pregnancy & Newborn Screening, Breast Screening, Cervical Screening, and For Health Professionals. The current page is titled "Newborn Metabolic Screening Programme - heel prick test". The page content includes a sidebar with links for "Antenatal HIV Screening", "Antenatal Screening for Down syndrome and other conditions", "Newborn Metabolic Screening Programme - heel prick test" (selected), and "Universal Newborn Hearing Screening Programme". The main content area features a header "Newborn Metabolic Screening Programme" with a sub-header "The Screening Process" and a photograph of babies. Below the photo, the text states: "The Newborn Metabolic Screening Programme screens for rare but potentially serious disorders such as phenylketonuria (PKU), cystic fibrosis, and congenital hypothyroidism. A blood sample is taken from your baby's heel at or as soon as possible after 48 hours of age (the 'heel prick' or 'Guthrie' test). If a disorder is found, early treatment can prevent permanent damage or death." A "Did you know that..." section lists: "While most babies look healthy, there are some disorders that aren't visible", "Early treatment of these disorders can prevent potentially serious complications which can cause permanent damage to the baby or even death", "To screen for these disorders, a sample of blood is collected from the baby's heel", and "The screening is free to babies born in New Zealand (<https://www.health.govt.nz/new-zealand-health-system/eligibility-publicly-funded-health-and-disability-services>)". A "How to get your baby screened" link is also present. The right sidebar contains sections for "The Screening Process" (with a "Find out how the screening process works" link), "Resources" (with a link to "Access information pamphlets and other resources"), and "Suggested links" (with links for "Eligibility for Publicly Funded Health and Disability Services" and "Frequently asked questions").

Parent information

- Available since 1980
- Revised at 5-7 year intervals
- Most recent revision November 2010 coordinated with development of policy framework and practitioner and parent DVDs



2005

- Public consultation on storage and use of residual dried blood spots
 - Focus groups
 - On-line surveys
 - Stakeholder workshops
- Expert advice
- Outcome
 - Destroy all immediately
 - Save all for every possible research
 - And everything in between

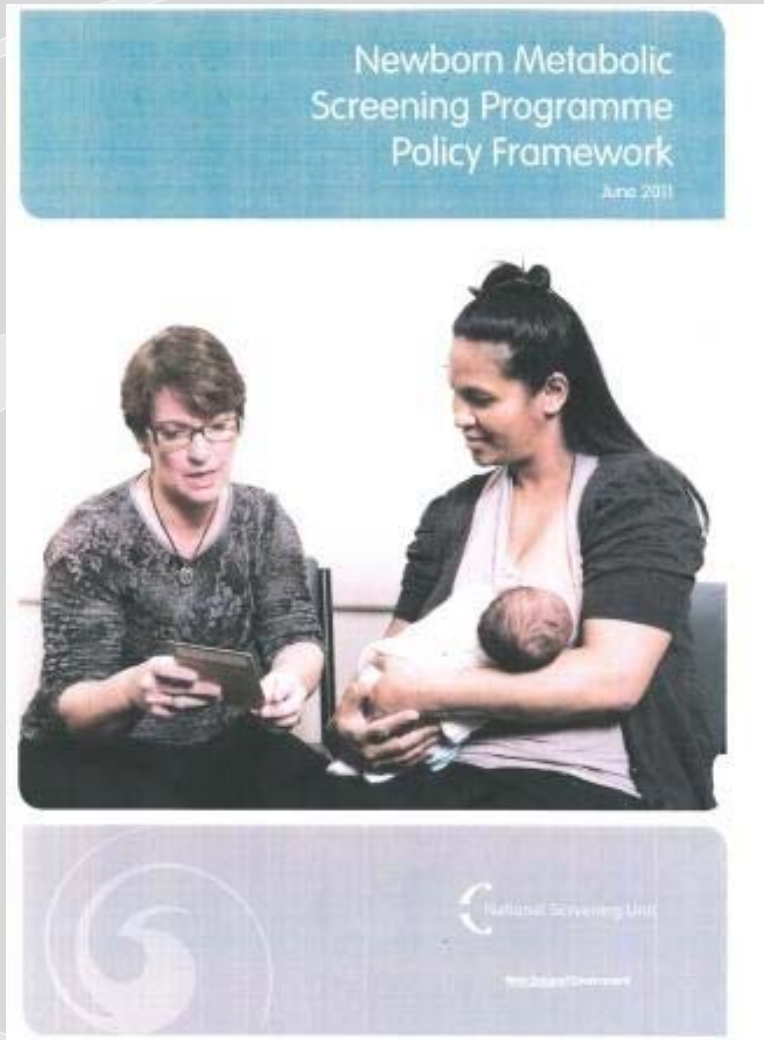
2006 -> 2011



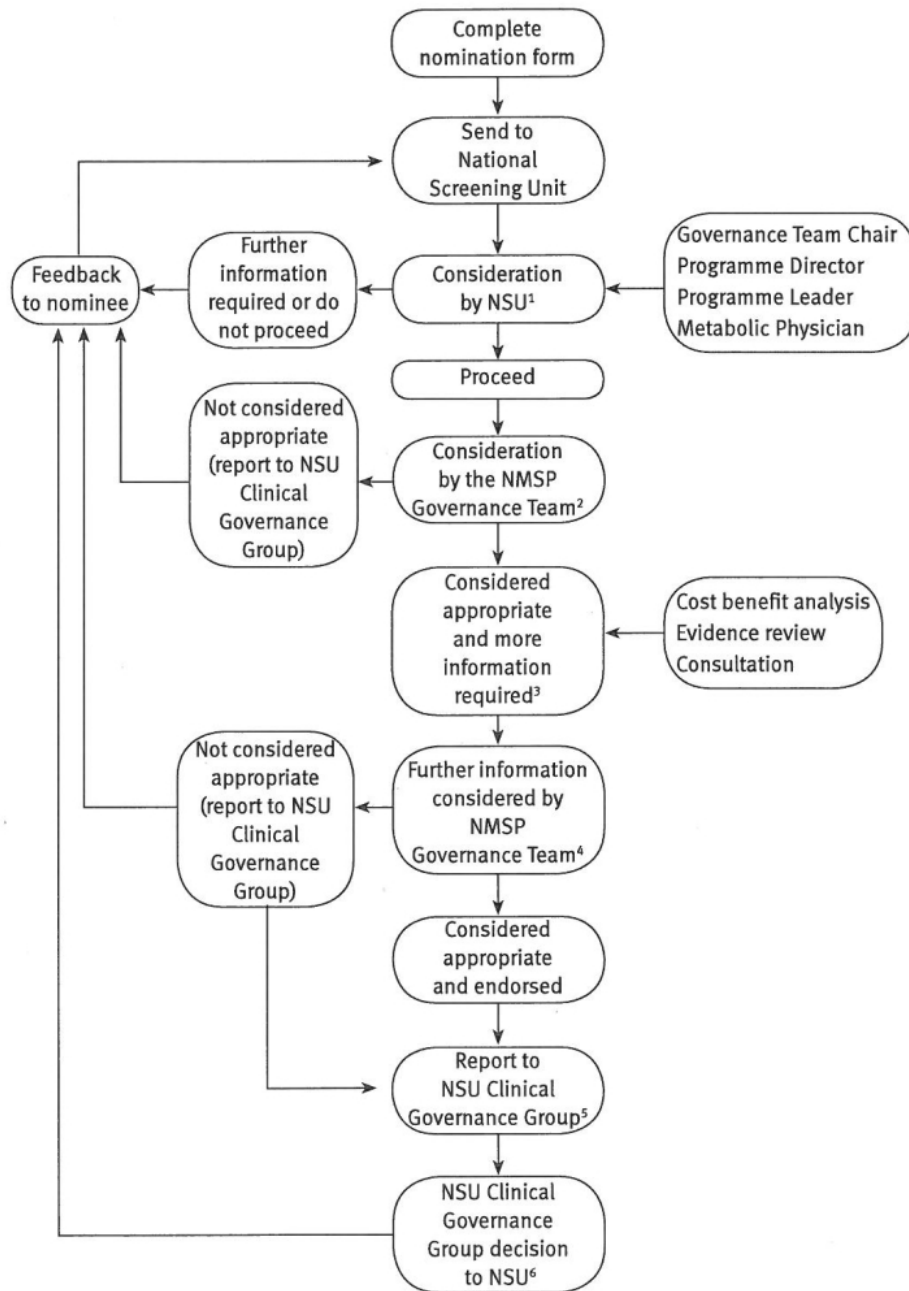
- Ministry of Health Options paper
- Sent to Cabinet Social Policy Committee
- Decided on permanent retention but
 - New Governance team for programme
 - New process for requesting access for research (approval of Governance team as well as Ethics Committee)
 - Research use of samples collected before June 2011 must have consent. After that time consent implied as to be more thoroughly covered in information sheet

Policy Framework

June 2011



- Return of residual spots to family
- Storage and use
 - Separate consents
 - Primary uses
 - Secondary uses
 - Family health uses
 - Police use – separate MOU
 - Coroner
 - Mortality review
 - Research uses



- Research application form
- Process flowchart

Informed consent

- Consent must be obtained
- Separate consent for collection and storage of samples
- Verbal consent sufficient if documented ideally in baby's notes, otherwise mother's

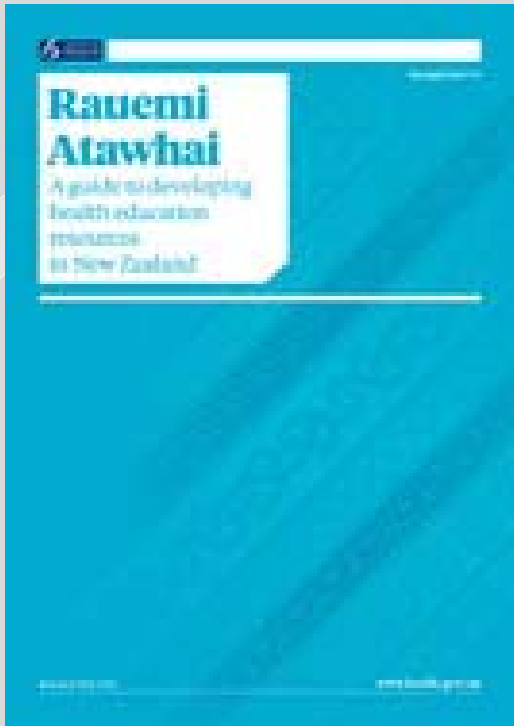
Primary uses

- Screening newborns
 - First test
 - Repeat confirmatory testing
 - Investigation of false +ve and –ve
 - Programme QA and audit
 - Assay improvement and test validation for current disorders
 - Test validation for potential new disorders

Secondary uses

- For the benefit of the baby and family eg congenital cmv
- Forensic / police investigations
- Coroner investigations
- Mortality review
- Research
- Other

Rauemi Atawhai



- MOH guide to developing health education resources in New Zealand
 - Be prepared
 - Be clear on your audience and your key messages. Language level
 - Be relationship focused
 - Test, test and test again with your audience and stakeholders.
 - Pictures not cartoons

Process

- Contract let to commercial company
- Text review, design development
- Pretesting – 40 people NZ European, Maori, Pacific, Asian – range of locations
 - Clarity
 - understanding of messages
 - Usefulness
 - cultural appropriateness
 - Relevance
 - Appeal
 - design

- NSU consultation with key stakeholders including consumers, members of the NMSP Advisory Group, professional bodies
- Completion of photo shoot for new photos for the pamphlet - required signed consent for use.
- Final review and editing
- Printed November 2010



Newborn Screening
Free health checks for your baby

Your newborn baby's blood test



The Newborn Metabolic
Screening Programme

All babies are checked at birth to see that all is well. Some of your baby's health checks are called 'screening'.



The Newborn Metabolic Screening Programme

The Newborn Metabolic Screening Programme detects rare but life-threatening metabolic disorders with a blood test done at 48 hours old or as soon as possible after this. Since 1969, almost all babies in New Zealand have had this screening. Early diagnosis means that treatment can start quickly, before the baby becomes sick. Metabolic disorders are hard to find without screening.

Why screen for metabolic disorders?

Screening saves lives. Each year, about 45 New Zealand babies are found to have a metabolic disorder. Although these disorders cannot be cured, early treatment with medication or a special diet can help your baby stay well and prevent severe disability or even death.

Metabolic disorders can occur in any family, even when there is no family history of disorders. Screening is an important way of identifying babies who are more likely than other babies to have a disorder.

Who is newborn metabolic screening for?

Newborn metabolic screening is offered free for babies born in New Zealand. Your midwife or doctor will talk with you during pregnancy about screening for your baby.

The Ministry of Health strongly recommends screening for your baby.



- About the programme
- Why screen?
- Who should be screened (not compulsory but strongly recommended by the Ministry of Health)

How is the blood sample collected?

The blood sample is collected from your baby's heel onto a blood spot card. The blood spot card is sent to the laboratory for testing. For the most accurate test results, the sample must be collected when your baby is 48 hours old or as soon as possible after this.



It is your decision to have your baby screened. When your baby has screening, you also need to decide whether the leftover blood spots are stored or returned to you after screening.

Test results

Your midwife or doctor will receive your baby's results and tell you what they are. This will be within 10 days if the results are negative. If any result is positive, the laboratory will notify your midwife or doctor as soon as possible.

Why is another blood sample sometimes needed?

Another blood sample may need to be taken if the first test result is not clear. If another sample is needed from your baby, your midwife or doctor will tell you why.

It is important that the new sample is taken as soon as possible so that your baby completes screening.

What if my baby has a positive result?

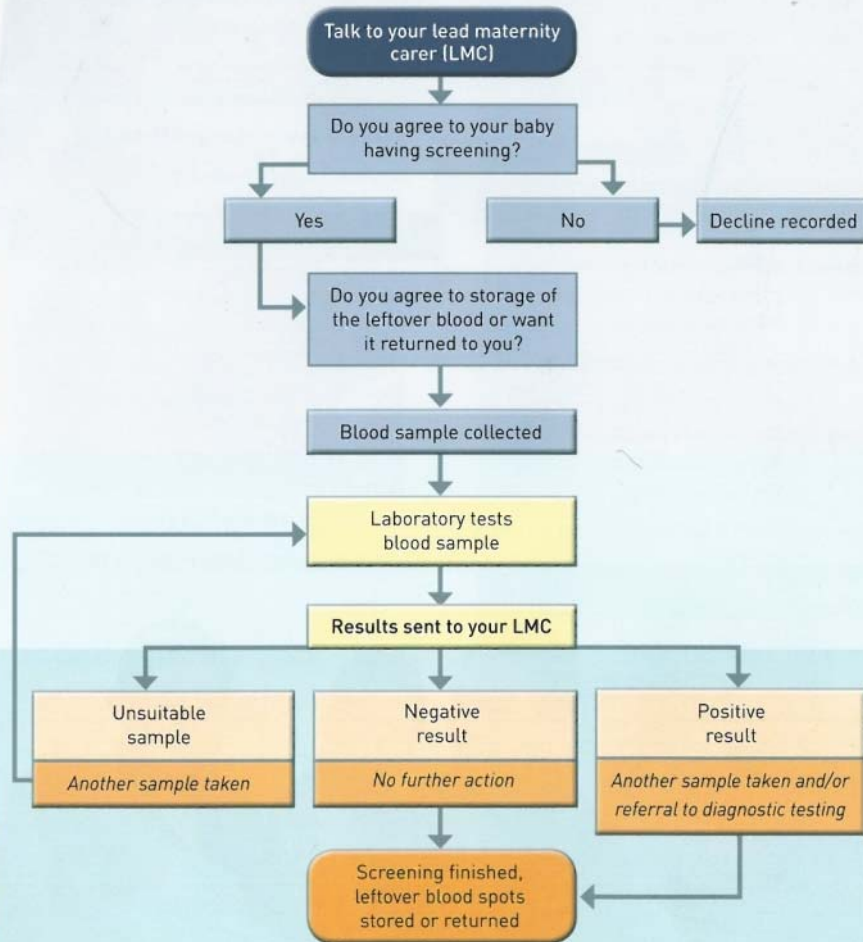
If your baby has a positive screening result, a further sample and/or referral to a specialist may be required. The specialist will examine your baby and order diagnostic testing to confirm whether your baby has a disorder or not. It is important to complete the diagnostic testing as soon as possible so that if your baby has a disorder, treatment can start straight away.

Screening identifies almost all babies who have a disorder. There is a small chance that some babies may be missed or a screening result will be positive for a baby who does not have a disorder.



- How is the sample collected?
- Can choose return of residual sample
- How results will be given
- Why are repeats needed?
- About positive results

The newborn metabolic screening process



- Screening process

What if my baby hasn't been screened?

If your baby is more than three days old and you don't think he or she has been screened, talk to your midwife or doctor.

Storage and use of leftover blood spots

Any blood left over after screening is either securely stored or returned to you. Blood is stored to assist with maintaining a high-quality screening programme and for other uses.

What can the stored blood spots be used for?

The stored blood spots may be used:

- for repeat testing. If your baby has a disorder but did not have a positive test result, the blood sample can be tested again to see why this happened.
- to improve the screening programme, such as by making sure that testing equipment produces accurate results
- for research approved by an ethics committee
- to investigate a death or illness in your family.

The stored blood spots will not be used for anything else without written consent from the parents or guardians or from another lawful authority, such as if ordered by a court. More information is available at www.nsu.govt.nz



How do I get leftover blood spots returned to me?

You can ask your midwife or doctor to arrange for the leftover blood spots to be returned to you by sending a signed request with the blood spot card. Alternatively, leftover blood spots can be requested at any time using the form 'Return of Newborn Metabolic Screening Samples' available at www.nsu.govt.nz



- What are stored spots used for?
 - Repeat testing
 - Screening lab uses
 - Research approved by an ethics committee
 - Family health reasons
- How do I get spots returned to me?

What information is collected and how is it used?

As part of newborn metabolic screening, basic information about your baby is collected and stored. This includes your baby's name and address, sex, ethnicity and weight, and where and when your baby was born. Your name is also recorded. The programme holds this information securely and confidentially.

The information is used to:

- interpret screening results
- check that babies have been screened
- make sure that results can be given to your midwife or doctor
- monitor the screening programme.

The Ministry of Health collects information for monitoring and evaluation of the screening programme. Your decisions about screening will be recorded in your maternity and Well Child Tamariki Ora notes. If you choose not to have your baby screened, you will also be asked if this information can be sent to the screening programme.

What are my rights?

The Code of Health and Disability Services Consumers' Rights protects your rights. You can read more about these rights at www.hdc.org.nz

The Health Information Privacy Code protects your privacy. You can read about the code at www.privacy.org.nz



What metabolic disorders are babies screened for?

Babies are screened for over 20 treatable disorders. A full list of the disorders is available at www.nsu.govt.nz

Amino acid disorders, eg, phenylketonuria (PKU)

Caused by a missing enzyme. Without these enzymes, amino acids (such as phenylalanine) rise to harmful levels

Can lead to brain damage and life-threatening complications

Treated by special diet

Occurs in about 5 babies every year

Fatty acid oxidation disorders, eg, medium chain acyl-Co A dehydrogenase (MCAD) deficiency

Caused by a missing enzyme. Without these enzymes, the body cannot break down fats to make energy

Can lead to life-threatening complications

Treated by ensuring regular feeding (a special diet is needed in some disorders)

Occurs in about 5 babies every year

- What information is collected?
- How is it stored and used?
- What are my rights?
- What conditions are screened for?
 - Amino acid disorders
 - Fatty acid oxidation disorders

Congenital hypothyroidism

Caused by not enough thyroid hormone

Can lead to slowed growth and developmental delay

Treated by Thyroxine

Occurs in about 20 babies every year

Cystic fibrosis (CF)

Caused by a defective gene and its protein product, leading to thick, sticky mucus

Can lead to poor growth, chest infections and shortened life

Treated by high-calorie diet, medicines and physiotherapy to keep the lungs healthy

Occurs in about 8 babies every year

Congenital adrenal hyperplasia (CAH)

Caused by lack of an enzyme in the adrenal gland

Can lead to life-threatening complications

Treated by steroid medication

Occurs in about 3 babies every year

Biotinidase deficiency

Caused by lack of an enzyme, leading to a deficiency of biotin

Can lead to life-threatening complications

Treated by taking vitamin H (biotin)

Occurs in about 1 baby every 3 years

Galactosaemia

Caused by an enzyme defect that prevents normal use of milk sugar

Can lead to jaundice, cataracts and life-threatening illness

Treated by special diet including replacement of milk-containing foods

Occurs in about 1 baby every 2 years



The Newborn Metabolic Screening Programme is committed to the highest possible standards. To maintain the quality of the programme, disorders screened for are reviewed and the programme is closely monitored. Further details are available at www.nsu.govt.nz

- What conditions are screened for
 - Congenital hypothyroidism
 - Biotinidase deficiency
 - Cystic Fibrosis
 - Galactosemia
 - Congenital adrenal hyperplasia
- Headings
 - Caused by
 - Can lead to
 - Treated by
 - Occurs in



More information

It is important that you have enough information to help you decide about newborn metabolic screening.

If you would like more information:

- ask your midwife or doctor
- check online at www.nsu.govt.nz

Your midwife or doctor can provide you with a DVD about newborn metabolic screening, or you can view this at www.nsu.govt.nz

The Newborn Metabolic Screening Programme is overseen by the National Screening Unit of the Ministry of Health.

This resource is available at www.healthed.govt.nz or the Authorised Provider at your local DHB.



New Zealand Government

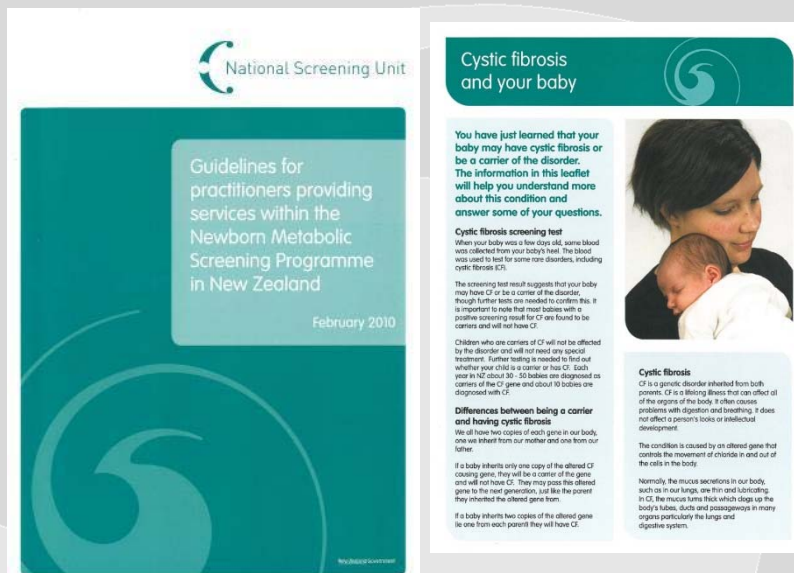
November 2010. 06/2012. Code HE2310

- How to get more information

Information sheet complemented by



- Best practice DVD for families
- Disorder positive information sheets



- For healthcare practitioners
 - Guidelines
 - On-line learning

Resource availability



- Your Newborn Baby's Blood Test
- On-line learning
- DVDs
- Guidelines
- Policy Framework
- Monitoring Framework
- Downloadable free from <http://www.nsu.govt.nz/current-nsu-programmes/newborn-metabolic-screening.aspx>
(national screening unit new zealand newborn)

Thank-you

