

Newborn Metabolic Screening Programme: Key Messages and Questions and Answers

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General Communication / Key Messages

- The Newborn Metabolic Screening Programme screens newborns at 48 hours of age for over 20 metabolic disorders. These disorders are rare and can be life threatening.
- Approximately 45 babies a year are identified with one of the disorders.
- Since 1969, almost all babies in New Zealand have had screening. Early diagnosis of one of the metabolic disorders means that treatment can be started quickly before the baby becomes sick.
- For blood spot cards collected after June 2011, parents will be properly informed of the potential primary and secondary uses of the blood spot card before consenting to storage of the cards.
- The screening information is provided and discussed with the parents prior to consent being obtained for the screening test and to make a decision regarding storage.
- Parents need to decide whether to a) request the return of the blood spot card after screening, or b) for it to be stored securely by the programme.
- Parents/guardians can request the return of their baby/child's blood spot card at any time up until the age of 16.
- Individuals aged 16 or older can request their blood spot card be returned to them at any time.
- Once screening is completed the blood spot cards are returned to the parent/guardian or stored indefinitely in secure storage.
- The stored blood spot samples may be used:
 - for repeat testing. If a baby has a disorder but did not have a positive screening result, the blood sample can be tested again to see why this happened
 - to improve the screening programme
 - to investigate a death or illness in a family
 - for victim identification, governed by a Memorandum of Understanding with the New Zealand Police
 - for research approved by an ethics committee

- Stored blood spot samples cannot be used for identification of criminals, unless there is a court order. This has never occurred in the history of the Programme.
- Blood spot samples stored prior to the policy implementation (pre June 2011), requires individual written consent if requested for population research.
- Any proposal for research that will require release of cards collected after June 2011 must first be approved by an Ethics Committee and is then reviewed by the Programme's Governance Team.
- In 2005 the National Screening Unit, within the Ministry of Health, became responsible for the national leadership of the Newborn Metabolic Screening Programme. Prior to this the Programme was managed by LabPlus, Auckland District Health Board.
- The National Screening Unit conducted public consultation, focus groups and stakeholder workshops during 2007-2009 on the Newborn Metabolic Screening Programme including the storage and use of the blood spot cards. Reports can be found at www.nsu.govt.nz.
- The Ministry of Health has taken into account the range of views expressed in the consultation processes and has also included expert advice in the *Policy Framework*.
- The Policy Framework formalises the policy for programme activities including the requirements for laboratories, LMCs, the programme Governance Team and the Ministry of Health.
- More information on the disorders and the screening programme can be found at www.nsu.govt.nz.

Questions and Answers

- **What is the Newborn Metabolic Screening Programme?**

The Newborn Metabolic Screening Programme detects rare and life-threatening metabolic disorders with a blood test done at 48 hours of age 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?**

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

- **Who is metabolic screening for?**

Newborn metabolic screening is offered free for babies born in New Zealand. The Ministry of Health strongly recommends metabolic screening.

- **How is the blood sample collected?**

The blood sample is collected by a health practitioner, usually a midwife, performing a heel prick to collect a sample of blood from a baby's heel onto a blood spot card. Once the blood is dry, the blood spot card is sent to the laboratory for testing. For the most accurate test results, samples are collected at 48 hours of age or as soon as possible after this.

- **What are the consent requirements for the programme?**

The midwife or doctor looking after the pregnant mother will provide information and discuss screening during pregnancy. A further conversation will take place once the baby is born and verbal consent is required. The consent and the discussion are recorded in the clinical notes.

A discussion is also required with the parents/guardians to decide whether to a) request the return of the blood spot sample after screening or b) for it to be stored securely by the programme.

- **How long have blood spot cards been stored?**

Blood spot cards have been stored since the programme began in 1969.

- **How do I get a blood spot returned?**

Parents/guardians can request the return of the blood spot card by asking the person who takes the heel prick to arrange that it is returned, by sending a signed request with the blood spot card.

Parents/guardians or individuals of age can request the return of blood spot cards at any time using the 'Return of newborn metabolic screening samples' form available on the NSU website www.nsu.govt.nz.

- **What part of the blood spot card gets returned to me?**

The blood spots themselves are returned to parents/guardians or individuals who request them. The programme recommends that the blood spots are kept by parents/guardians or individuals in a safe dry place.

- **What happens to the blood spots after testing?**

For most blood spot cards, there is blood left over after screening is completed. Once screening is completed, the blood spot card is either returned to the parents/guardians or individual if requested or stored indefinitely in a secure locked area. Parents/guardians or individuals can request the return of their blood spots at any time, using the 'Return of newborn metabolic screening samples' form available on the NSU website www.nsu.govt.nz.

Only authorised staff from the screening programme can access stored blood spots.

- **What data is collected and how is it stored?**

When a heel prick is performed, information is provided on the card. This information includes demographic data (for example the baby's name, sex, birth date, time, weight, the mother's details and LMC details). These details are entered into a secure information system. Once screening is completed, the results of the screening tests are also captured in the information system. This data is used for screening purposes and is only accessed by authorised personnel. The Ministry of Health also collects information for monitoring and evaluation of the screening programme.

- **What are stored blood spots used for?**

Stored blood spots may be used for:

- for repeat testing. If a baby has a disorder but did not have a positive screening result, the blood sample can be tested again to see why this happened
- to improve the screening programme
- to investigate a death or illness in a family
- for victim identification, governed by a Memorandum of Understanding with the New Zealand Police
- for research approved by an ethics committee.

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- **How often have blood spots been used for research?**

To date, blood spot cards have not been used for large scale population studies.

- **Is DNA testing carried out on the blood spots?**

As part of metabolic screening, about 1 percent of samples are currently tested for a change in DNA (mutation) associated with cystic fibrosis. No other DNA testing is done on samples unless authorised by the parents/guardians/individual or through legal avenues, such as a court order.

- **What is the process for research on blood spot cards?**

The Policy Framework sets out the requirements for population research studies. These include:

- The study must have ethics committee approval
- The study must have Ministry of Health approval
- The study must be presented to the Programme Governance Team
- The research may not use up all the blood on an individuals card
- The research must be considered an appropriate use of residual blood spot samples and contribute to the public good through increased scientific knowledge.