NEWBORN BLOODSPOT SCREENING

THE SAFETY OF THE SCREENING PROCESS
Screening is quick and safe. Because the heel prick causes a small break in the skin, there is a small risk of infection. The midwife or nurse collecting the sample will use gloves and clean the heel before the test. You may wish to breastfeed or hold your baby during the test as the heel prick may cause a little discomfort to your baby.

 Babies to be screened
Parents consent for screening
Midwife/nurse does screening
Card sent to laboratory for testing
Result negative
Storage Min 2 years - 18 years
Result positive
Midwife/nurse arranges retesting
Result positive
Referred to specialist
Result negative

WHAT IS A NEGATIVE RESULT?
A negative screening result means your baby has not been identified as having one of the conditions being screened. On very rare occasions, a false negative may occur where the baby has a normal screen but develops symptoms for one of the conditions later. This occurs in approximately one in 100,000 cases.

WHAT IS A POSITIVE RESULT?
A positive screening result does not necessarily mean your baby has a particular condition. Newborn screening identifies babies at increased risk of a condition. Further testing is needed to confirm the result. Babies with a positive screening result have more samples collected. If the later screens are also positive, your baby will be referred to a specialist.

DNA TESTING
Newborn bloodspot screening involves biochemical testing not DNA testing. However, approximately one per cent of babies from the biochemical testing will show a risk for cystic fibrosis or a fatty acid oxidation disorder which will then be DNA tested. No DNA tests are done on any other samples.

HAVING A LATER SCREENING
If you choose not to have your baby screened and you change your mind later, speak to your family doctor. Your doctor will arrange for your baby to have the right tests.

MORE INFORMATION
For more information on the NSW Newborn Screening Program:
• Visit the program website
health/statewide-laboratory-services/
nsw-newborn-screening-programme

MORE ABOUT SCREENING

73 Miller Street, North Sydney, NSW 2060
Locked Mail Bag 961, North Sydney 2059
Tel: 61-2-9391 9000
www.health.nsw.gov.au

IMPORTANT Please keep this information for three months after your baby is born. You may receive a request to have the test repeated or your doctor or midwife may need to follow up your baby’s test results with further investigations.
Newborn Bloodspot Screening is a free blood test that is offered to every newborn baby in NSW and the ACT. This blood test is to check if your baby may have been born with a medical condition.

If your baby is not screened at birth and has one of the conditions being screened, the baby’s development may already be affected by the time symptoms appear. Some of the conditions that are screened for may be life threatening if treatment is delayed. Early screening and detection means treatment can start early.

If you decide not to have your baby screened

If you choose not to have your baby screened, you will be asked to sign a separate refusal of screening form. Your baby’s screening card will still be sent to the laboratory with the completed refusal form. If you decide not to have your baby screened, we recommend that you let your family doctor and your child and family health nurse know that your baby has not been screened.

After screening

If the screening results are normal, you will not be contacted.

If a repeat screening is needed, your midwife or child and family health nurse will arrange it. There are a number of reasons why you may be contacted:

1. The laboratory may need to have a repeat blood sample.
2. If you are notified that your baby has an abnormal screening result, more samples will be collected and tested.
3. If you are notified that your baby has an abnormal screening result, you will be referred to a specialist.

Storage of screening cards

Screening cards are stored in a secure, locked area at all times. Access to stored cards is tightly controlled and protected by state legislation.

The laboratory will keep your baby’s screening card for two years for quality assurance and audit purposes. After two years, you can ask for your baby’s screening card to be returned to you or destroyed if you no longer want it stored. If you are happy for the screening card to be stored, it will be retained by the laboratory for 16 years. The legal age of consent is 18 years so once your child turns 18 the card is then destroyed as they did not consent to the test.

Who can access the card

The Newborn Bloodspot Screening Program screens approximately 100,000 babies per year for 25 medical conditions. Only a small number of babies will be diagnosed with one of the medical conditions of which the following are the most common.

Primary congenital hypothyroidism
- An absence or abnormal formation or function of the thyroid gland affects growth and causes intellectual disability if untreated.
- Around 40 babies per year are diagnosed.
- Treatment is to medicate with thyroid hormone.

Cystic Fibrosis
- A dysfunctional gene results in thick mucus in different organs throughout the body, which leads to severe chest infections and a failure to thrive if untreated.
- About 30 babies per year are diagnosed.
- Individuals with cystic fibrosis have a great improvement in their health if they start treatment early.

Phenylketonuria (PKU)
- The body is unable to break down the essential amino acid phenylalanine and can lead to severe intellectual disability if untreated.
- About 10 babies per year are diagnosed.
- The treatment for PKU is a diet low in phenylalanine started in the first two to three weeks of life.

Medium Chain Acyl CoA Dehydrogenase (MCAD)
- Inability of the body to completely break down fat. If untreated, it may be life-threatening during common childhood illnesses.
- About 6-8 babies a year are diagnosed.
- Treatment involves taking extra precautions during illnesses to ensure the child gets adequate energy intake.

Galactosaemia
- The body is unable to process galactose and can lead to liver failure and other infections.
- About 1-3 cases per year are diagnosed.
- Treatment is a galactose-free diet commenced before 2 weeks of age.

Other rare metabolic disorders
- There are many other rare metabolic disorders, including disorders of the metabolism of amino acids, urea cycle, organic acids and fatty acid oxidation.
- Many can lead to severe disability or death.
- A total of around 20 babies per year are diagnosed.