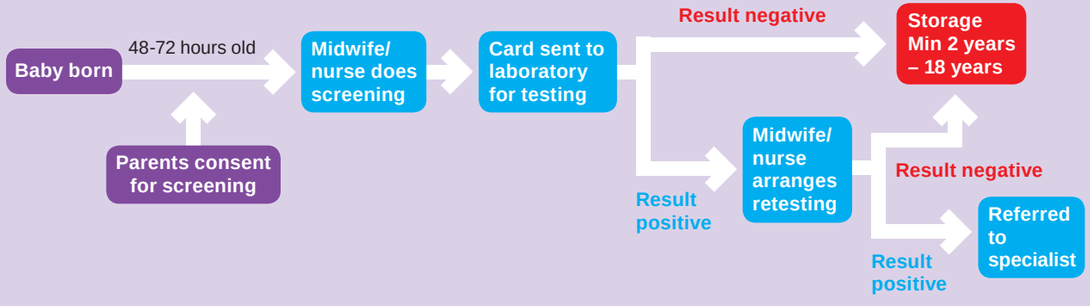
MORE ABOUT SCREENING

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.

The Newborn Bloodspot Screening Process



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?

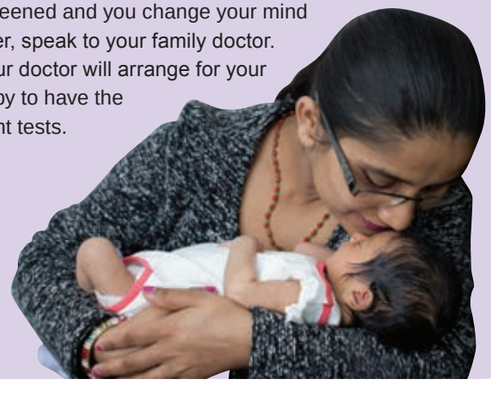
A positive screening result does not necessarily mean your baby has a particular condition. Newborn screening identifies babies at increased 'risk' of a condition. Further testing is needed to confirm the result. Babies with a positive screening result have more samples collected. If the later screens 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:

- Watch the video at http://www.kidsfamilies.health.nsw.gov.au/3341.aspx
- Visit the program website
- http://www.schn.health.nsw.gov.au/professionals health/statewide-laboratory-services/ nsw-newborn-screening-programme

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.

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SHPN: (NFK) 150441 May 2016

NEWBORN BLOODSPOT SCREENING

TESTS TO PROTECT YOUR BABY

EARLY DIAGNOSIS IS IMPORTANT TO YOUR BABY'S FUTURE HEALTH AND WELLBEING

NSW GOVERNMENT Health

WHY SCREENING IS IMPORTANT

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. While a baby will be offered treatment when symptoms appear, their growth and development could already be affected. 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.***

When Screening Occurs

When your baby is between two and three days old (that is between 48 and 72 hours old), your baby will have the test. A midwife or nurse will prick your baby's heel with a lancet (a special needle) and will collect a few drops of blood onto a screening card. Then, the card is sent to a central laboratory for processing.

CONSENT FOR SCREENING

Before a sample is collected, you must give NSW Health your signed consent that you agree to the screening. There is a consent section on the screening card (see below) which you will be asked to sign if you want to go ahead with the screening.

Consent on the screening card

NSW NEWBORN SCREENING PROGRAMME

Consent for Collection and Testing of Sample

I have received and understood the information in the NSW Newborn Screening pamphlet.

I consent to my baby having blood collected and tested Yes [ ] No [ ]

Storage of screening card for greater than 2 years

I consent to the storage of the screening card for longer than 2 years Yes [ ] No [ ]

I understand that blood from screening cards may be used for de-identified health research.

I agree to make my baby's blood sample available for this purpose Yes [ ] No [ ]

Cards without consent will not be used for research

Parents signature:

Sign here

Tick 'Yes' or 'No':

1. Consent to have your baby screened.
2. Consent to store the screening card for more than 2 years.
3. Consent for the card to be used for health research if personal information is removed.

You can choose to say YES to the first and NO to the other two items if you wish.

If you choose 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.

If after further testing your baby's blood sample remains positive for one of the conditions, 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 18 years. The legal age of consent is 18 years old so once your child turns 18 the card is then destroyed as they did not consent to the test.

Who Can Access the Card

Your baby's card will only be accessed:

- If further clinical testing is recommended for your baby
- By the laboratory for quality control and audit purposes
- For ethics approved, health research where all personal details are removed so your baby cannot be identified
- By a Court order
- By the Coroner.

SCREENING FOR PARTICULAR MEDICAL CONDITIONS

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) Deficiency:

- 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.