NSW Health

Newborn bloodspot screening

Tests to protect your baby

Early diagnosis is important to your baby's future health and wellbeing.

NSW GOVERNMENT

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. Newborn bloodspot screening involves a combination of biochemical and genetic testing.

Consent for screening

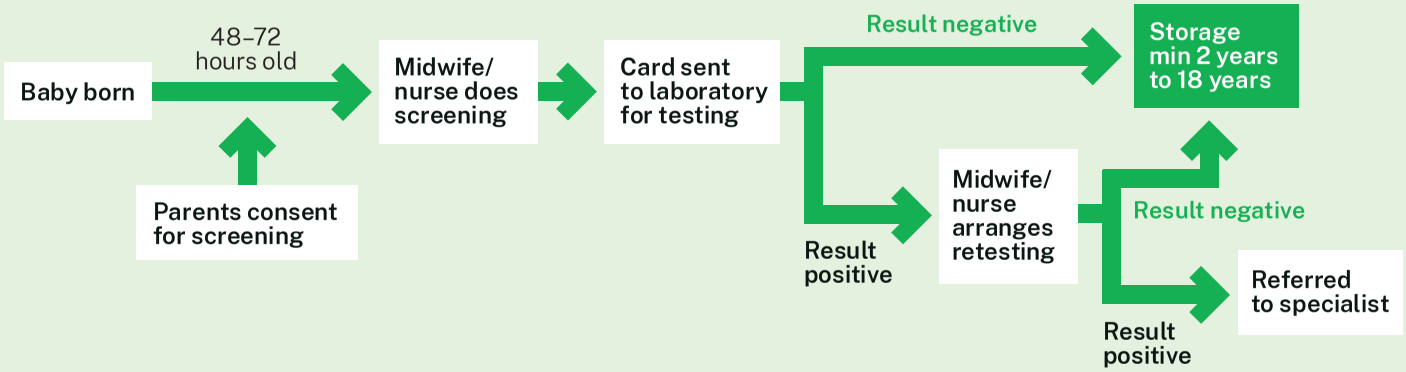
Before a sample is collected, you must give NSW Health your signed consent that you agree to the screening. There is a consent section on the screening card (see next page) which you will be asked to sign if you want to go ahead with the screening.

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



Consent on the screening card

Consent for scoping studies

From time to time, the program may trial screening for additional conditions. When this happens, there will be an extra tick box for you to consent into these scoping studies.

NSW Newborn Bloodspot Screening Program

Consent for collection and testing of sample	Yes	No
I have received and understood the information in the NSW Newborn Screening pamphlet.	☐	☐
I consent to my baby having blood collected and tested.	☐	☐
Storage of screening card for greater than 2 years		
I consent to the storage of the screening card for longer than 2 years.	☐	☐
I agree to make my baby's blood sample available for de-identified health research.	☐	☐

Cards without consent will not be used for research.

Tick 'Yes' or 'No':

1. Acknowledgment that you **have** been given the NSW newborn screening brochure and understand it.
2. Consent to have your baby screened.
3. Consent to store the screening card for more than 2 years.
4. Consent for the card to be used for de-idenitfied health research and all personal information is removed.

You can choose to say YES to the first two and NO to the remaining 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.

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.

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.

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, then an additional sample will be collected for further testing.

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 a maximum period of 18 years.

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.

Approximately 1 in 800 babies will be identified with one of the conditions

The most commonly diagnosed conditions are:

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.

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.

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.

Congenital Adrenal Hyperplasia

- Altered ability of the adrenal gland to produce hormones that may affect the baby's metabolism, response to infection, ability to regulate salt levels and sex characteristics.
- About 6-7 babies a year are diagnosed.
- The treatment is through medication such as hormone replacement and salt replacement.

Spinal Muscular Atrophy (SMA) and Severe Combined Immunodeficiency (SCID)

- In SMA the body is unable to make a specific protein called SMN that is required for nerve formation.
- Treatment is by giving the baby this SMN protein by a complex medical procedure.
- In SCID the baby has a poor immune system that is unable to fight infections.
- Treatment is to provide specific medical treatment to overcome the poor immune system.

Further conditions may be added or removed from the screening program.

Duchenne Muscular Dystrophy
Scoping Study

A scoping study is underway in NSW to check if newborns should be screened for Duchenne muscular dystrophy (DMD).

What is a scoping study?

In a scoping study, a health condition is temporarily added to the newborn bloodspot screening program.

Understanding Duchenne muscular dystrophy (DMD)

DMD is a genetic condition affecting boys. DMD affects the way a boy can move with time and usually difficulties with movement start to appear in childhood. Each boy with DMD is impacted differently. There's currently no cure, but medicines and therapy can help manage symptoms and slow its progress.

Why would I say yes to participating in the scoping study for DMD?

At the moment, conditions are included in newborn bloodspot screening programs if there's a proven medical treatment available early on. However, for this DMD scoping study, there is no proven treatment required in the first years of life. Yet, ongoing research might lead to new treatments or clinical trials in the next few years. A positive result on newborn screening could mean an earlier diagnosis, and the possibility to access clinical trials and treatments earlier.

Details about the scoping study

This study has been approved by Sydney Children's Hospitals Network HREC (Approval number 2023/ETH02489).

Why would I say no to participating in the scoping study for DMD?

For some families, knowing about a medical condition in the newborn period, before their child shows symptoms, can be distressing, especially if there is no immediate action or medical treatment.

What does participation involve?

If you would like to participate in the scoping study, tick the 'yes' box on the back of your child's dried bloodspot card. For more information refer to the Consent section of this booklet.

The NSW Newborn Bloodspot Screening Program will then check if the permission is given for the new test to be performed on your child's dried bloodspot card. Nothing further will be required from you and your child to participate. As usual, if the results are normal, you will not hear anything from the screening program. For more information on the screening process, please see The Newborn Bloodspot Screening Process of the brochure.

Who is organising the DMD Scoping Study

This study is led by the Sydney Children's Hospitals Network (SCHN) Chief Investigator Clinical Professor Bruce Bennetts, Head of Department of Molecular Genetics at the Children's Hospital at Westmead.

Information on the Duchenne muscular dystrophy scoping study has been provided by Neuromuscular Clinical Specialists and the Genetic Counselling team under the TRAIL study (MRF2017165).

For more information visit our website

Thank you for your participation and for taking the time to read this information.

Have a question?

Email us at: schn-trail@health.nsw.gov.au

In partnership with:

Kids Research is part of Sydney Children's Hospital Network. The TRAIL study is supported by funding from the Australian Government under the Medical Research Future Fund.

More information

For more information on the NSW Newborn Screening Program visit the program website and watch the video at www.schn.health.nsw.gov.au/find-a-service/laboratory-services/newborn-screening or Sydney Children's Hospital Network website, www.schn.health.nsw.gov.au select 'Find a service', and go to 'NSW Newborn Screening Program'.

Newborn bloodspot screening program

Address: Locked Bag 2012 Wentworthville NSW 2145
Telephone: +61 278 253 659
Email: NSWH-newbornscreening@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.

Order additional newborn bloodspot screening brochures through your local brochure ordering system.

SHPN (HSP) 190744.

health.nsw.gov.au