Newborn bloodspot screening

Summary

- Every newborn baby in Australia is offered a test for rare, but serious, medical conditions.
- The conditions tested for are phenylketonuria, hypothyroidism and cystic fibrosis.
- You can choose whether you want your baby to have this test.
- A midwife will take a blood sample by pricking your baby’s heel. A few drops are collected on a piece of card.
- This usually happens between 48 and 72 hours after your baby is born.
- The card is then sent to a laboratory for testing.
- Most babies will have a normal test result.
- Sometimes a repeat test is needed. The results will usually be normal.
- If your baby is found to have a serious medical condition, you will be contacted to arrange treatment for your baby.

What is newborn screening?

Every newborn baby in Australia is offered a test for rare, but serious, medical conditions. This is called newborn screening.

The conditions tested for include:

- phenylketonuria (PKU)
- hypothyroidism
- cystic fibrosis

These conditions do not show any symptoms at birth and usually there is no family history.

It is very helpful to test for these conditions early, as treating them as soon as possible can prevent further health problems (complications).

Consent for newborn bloodspot screening

A midwife will offer you the newborn screening test in the hospital where your baby was born, or at home. If you want your baby to have the test, you will need to agree to the test in writing. This is called giving your ‘informed written consent’.

Declining newborn bloodspot screening for your baby

If you do not want your baby to have the test (this is called declining the test), you will need to sign a written statement to show that you understand the risks of not having it.

The conditions being screened for are rare. But, if your baby has one of these conditions and it is not discovered or treated, they could become permanently disabled or even die.

By the time symptoms of a condition do show, it may already have affected your baby’s development. Screening helps affected babies to be identified early. In most cases they can have treatment to reduce the health impact of the condition.

What happens when my baby is tested?
Usually, a midwife takes a blood sample between 48 and 72 hours after the baby is born. The midwife pricks the baby’s heel and places a few drops of blood onto a piece of card. This can happen in the hospital or at home.

The test is safe and will not harm your baby. The heel prick may cause brief discomfort to your baby, but holding or feeding them while the sample is collected may help. There is a small risk of infection, but the midwife will wear gloves and clean your baby’s heel before collecting the blood to make this risk as small as possible.

The blood sample is looked at in a laboratory. It is tested for different chemicals that show that a condition is present. These are called markers. If the marker for cystic fibrosis is found, the sample will be tested for the gene for cystic fibrosis.

Most babies will have a normal test result. You will not be contacted if the result is normal. Sometimes another test (a repeat test) is needed. Don’t worry if this happens, as most repeat screening results are normal.

If the test shows that your baby has a rare but serious medical condition, you will be contacted immediately to arrange treatment for your baby.

**Repeat tests and positive results**

Sometimes a repeat test is needed if the first sample:

- was collected too early
- was contaminated
- produced an unclear result.

Don’t worry if this happens, as most repeat screening results are normal.

If your baby is found to have a condition (this is known as a ‘positive screening result’), you will be contacted and referred to a specialist for further testing.

Positive screening results are usually confirmed by testing a sample of urine or blood.

**Conditions detected by newborn bloodspot screening**

Some of the conditions that can be detected by newborn bloodspot screening include:

- **phenylketonuria (PKU)** – a condition where the body cannot break down phenylalanine (found in food proteins). If untreated, PKU can lead to intellectual disability
- **hypothyroidism** – a condition where the thyroid gland does not make enough hormones. This can cause severe intellectual disability and growth problems. Early detection and treatment helps children to be healthy
- **cystic fibrosis** – a condition where organs such as the lungs and pancreas make abnormal mucus that clogs the organs and stops them working properly
- other rare conditions that affect metabolism – there are many other rare conditions that make the body unable to break down proteins and fats in the usual way. If these conditions are found and treated early before your baby becomes sick, they will have a better chance of having an improved health outcome.

**How newborn bloodspot screening is performed**

Blood is taken by pricking the baby’s heel. Having the heel prick may cause brief discomfort to your baby, but holding or feeding your newborn during the sample collection may help. Levels of biochemical markers for the above disorders are measured in the blood spots. The only genetic test performed is on a small number of samples (about one per cent) that have an increased level of the marker for cystic fibrosis.

**Repeat samples and positive results**

Sometimes, the laboratory may need a repeat blood sample if the first sample:

- Was collected too early
- Was contaminated
- Produced an unclear result.

Don’t be concerned if a repeat sample is required, as most repeat screening results are normal.
If your baby has a positive screening result (shows evidence of a condition), you will be contacted and referred to a specialist for further testing. Positive screens are usually confirmed by testing a sample of urine or blood.

**After newborn screening**

The screening card containing your baby’s blood sample will be stored in the laboratory for around two years. This happens in case more testing is needed and to help the laboratory make sure they are meeting quality standards.

You can also choose to make your baby’s screening card available for de-identified health research. An example of this type of research would be establishing normal values for a new newborn bloodspot screening test. All such projects require approval by an ethics committee.

After two years, cards are securely stored indefinitely. Access to stored cards is tightly controlled and protected by state legislation.

**Support is available**

The [Genetic Support Network Victoria (GSNV)](https://www.gsnv.org.au) supports people and families affected by genetic conditions. GSNV can help you to get the information, support and services you need.

If your child been diagnosed with a genetic condition, it may be helpful to speak to a genetic counsellor. Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand a condition and what causes it, how it is inherited (if it is), and what a diagnosis means for your child’s health and development, and for your family.

Genetic counsellors are trained provide information and support that is sensitive to your family circumstances, culture and beliefs.

**Where to get help**

- Your [GP (doctor)](https://www.gp.ca.gov.au)
- A [genetic counsellor](https://www.geneticcounsel.org)
- [Paediatrician](https://www.pediatrics.org)
- [Victorian Clinical Genetics Services](https://www.vcg.org.au) Tel. 1300 118 247
- [Genetic Support Network of Victoria](https://www.gsnv.org.au) Tel. (03) 8341 6315

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