Newborn bloodspot screening

Summary

- Every newborn baby in Australia is offered a newborn bloodspot screening test to identify those at risk for rare, but serious, medical conditions including PKU, hypothyroidism and cystic fibrosis.
- The blood is taken by pricking the baby’s heel.
- Most babies will have a normal screening test result.
- Occasionally, a repeat test is needed. The results will usually be in the normal range.
- If a baby is confirmed to have one of these conditions, the parents will be contacted to arrange prompt treatment for their baby.

Every newborn baby in Australia has the opportunity to undergo newborn bloodspot screening to identify those at risk of rare, but serious, medical conditions including phenylketonuria, hypothyroidism and cystic fibrosis. The benefit of this test is early detection – many complications of these disorders can be avoided with prompt treatment.

Usually, a blood sample is taken by a midwife between 48 and 72 hours after birth. The blood is taken by pricking the baby’s heel and placing a few drops of blood onto a screening card. This can take place in the hospital or at home. The test is safe and will not harm your baby. As with any procedure that breaks the skin, there is a small risk of infection, but this is minimised by using gloves and cleaning the heel prior to collection.)

The blood sample is examined in a laboratory. Most babies will have a normal screening test result. Parents are not contacted if the result is normal. Occasionally, a repeat test is needed. This should not cause concern, as most repeat screening results are normal.

If a baby is confirmed as having a rare but serious medical condition, the parents will be contacted immediately to arrange treatment for their baby.

Consent for newborn bloodspot screening

The hospital where your baby is born, or the midwife responsible for your care at home, will make sure you are offered the newborn bloodspot screening test for your baby, either during your hospital stay or at home. You will need to give your informed written consent before the test is performed.

Declining newborn bloodspot screening for your baby

If you decline the test for your baby, you will be required to sign a written statement to indicate that you understand the potential risks.

Parents should realise that, although the disorders being screened for are rare, if left unscreened and untreated, they could lead to permanent disability or even death for your child. These conditions do not show any symptoms at birth and usually there is no family history.

By the time symptoms of a condition do show, your baby’s development may already be impaired. Through screening, affected babies can be identified early and, in most cases, treated to prevent or minimise the health impact of the condition.

Conditions detected by newborn bloodspot screening

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

- Phenylketonuria (PKU) – a disorder where the liver does not produce enough of a particular enzyme and cannot metabolise (break down) phenylalanine, which is one of the 20 amino acid building blocks of protein.
untreated, PKU can lead to intellectual disability

- Hypothyroidism – a condition where the thyroid gland is underactive and fails to make enough hormones. The thyroid gland helps regulate metabolism. A lack of thyroid hormone can cause severe intellectual disability and growth problems. Early detection and treatment enables children to be healthy
- Cystic fibrosis – a condition where organs such as the lungs and pancreas secrete abnormal mucus that clogs the affected organs and stops them working properly
- Other rare metabolic disorders – there are many other rare metabolic disorders caused by faults in the breakdown of proteins and fat. Early detection and treatment, before babies become sick, results in a better 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 newborn, 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 bloodspot 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) supports people affected by genetic conditions by helping them to get appropriate and accurate information and support.

**Where to get help**

- Your doctor
- Paediatrician
- **Victorian Clinical Genetics Services** Tel. (03) 8341 6201
- **Genetic Support Network of Victoria** Tel. (03) 8341 6315

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