

Newborn screening

Every newborn baby in Australia undergoes newborn screening (NBS) to identify those at risk of rare, but serious medical conditions including phenylketonuria (PKU), 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 between 48 and 72 hours after birth by a midwife. The blood is taken by pricking the baby's heel and placing a few drops of blood onto a screening card. Once dried, the blood sample is examined in a laboratory.

The vast majority of 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.

Screening can detect some conditions

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

- **Phenylketonuria (PKU)** – a disorder where the liver does not produce enough of a particular enzyme and cannot metabolise (breakdown) phenylalanine – one of the 20 amino acid building blocks of protein. If untreated, PKU can lead to intellectual disability.
- **Hypothyroidism** – 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** – organs such as the lungs and pancreas secrete an 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 testing is performed

The blood is taken by pricking the baby's heel. 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 a repeat blood sample may be needed by the laboratory. This may be required if the first:

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

Don't be concerned 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.

After the screening

The screening card, containing the 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. 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 of Victoria (GSNV) wants all people with a genetic disorder in Victoria to have appropriate and accurate information and support. This is to enable them to manage the challenges to their health and wellbeing.

Where to get help

- Your doctor
- Paediatrician
- Genetic Health Services Victoria, Royal Children's Hospital Tel. (03) 8341 6201
- Genetic Support Network of Victoria Tel. (03) 8341 6315

Things to remember

- Every newborn baby in Australia is offered a newborn screening test to identify those at risk for rare, but serious, medical conditions including phenylketonuria (PKU), hypothyroidism and cystic fibrosis.
- The blood is taken by pricking the baby's heel.
- The vast majority of 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.

This page has been produced in consultation with, and approved by:

Genetic Health Services Victoria

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