Newborn bloodspot screening

For the health of your baby

What is newborn bloodspot screening?

Newborn bloodspot screening is a program that identifies babies at risk of having rare, but serious medical conditions that can affect normal development. The screening test is quick and safe and available for all newborns.

Screening is important because:

- Affected babies may not show any signs or symptoms of illness at birth.
- It allows the conditions to be identified early. With early detection, the conditions can be treated or managed in most cases.
- There are no alternative ways to identify babies with these conditions. By the time symptoms appear, their development may already be impaired.

Fortunately, most babies born in Victoria each year are healthy. Only a small number will be found to have one of these serious conditions. In some cases, the condition may be life threatening if treatment is delayed. In rare cases, the condition may be untreatable.

When and how is screening done?

Between 36-72 hours after birth, your midwife will collect a few drops of blood onto a screening card by pricking your baby's heel. If you are discharged early, the sample will be collected during a home visit.

Before a sample is collected, you must give your consent and sign the screening card. If you choose not to have your baby screened, you will also be asked to sign a separate 'decline of screening' form.

Samples for bloodspot screening are sent to the screening laboratory based at The Royal Children's Hospital in Melbourne.

Results

In over 99 per cent of cases, the results are normal. When this happens, parents are not contacted; final results for a small number of babies may take up to 6 weeks.

If your baby has an abnormal screening result, you will be contacted and referred to a specialist for further testing.

Sometimes, a repeat blood sample may be needed by the laboratory. This can happen for a number of reasons. Your midwife will arrange for a re-collection. Most repeat results are normal.

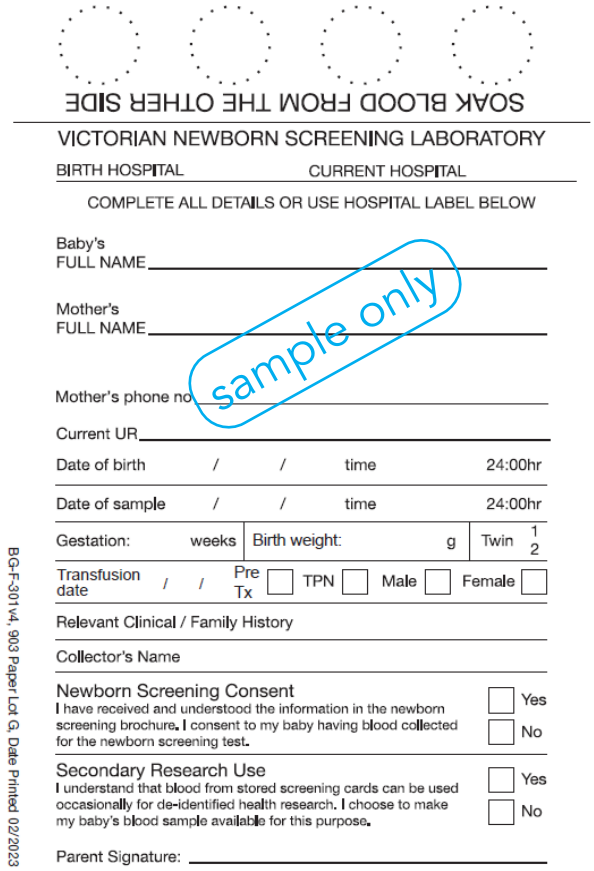
What does bloodspot screening detect?

The screening test covers around 26 different metabolic conditions. The following table gives information about the most common ones.

Disorder	Caused by	Problems if untreated	Treatment/ management
congenital hypothyroidism	thyroid gland unable to produce thyroid hormone (T3 & T4)	growth failure, intellectual impairment	thyroid hormone supplements
cystic fibrosis	abnormal secretions in the body; in particular the lungs & pancreas	impaired digestive & respiratory function, infections & decreased life span	dietary supplements, physiotherapy, medication
amino acid disorders* (e.g. phenylketonuria PKU)	defective enzymes that break down protein	developmental delay, intellectual impairment, seizures	dietary modifications, vitamin supplements
fatty acid oxidation disorders*	defective enzymes that turn fat into energy	muscle problems, poor feeding, vomiting, seizures, sudden death	avoid prolonged fasting, dietary modifications
congenital adrenal hyperplasia (CAH)	defective enzymes that help balance sugar and salt	increased male sex hormones, loss of water and salt in the urine, adrenal crisis, risk of sudden death	adrenal hormone supplements

* These disorders affect the breakdown of fat & protein in the body

The bloodspot screening card looks like this



Frequently asked questions

We have no family history and my baby seems healthy so why should they be screened?

The conditions screened for, as part of the newborn bloodspot screening program, usually do not show any signs or symptoms at birth and often there is no family history. By the time symptoms of a condition do show, 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.

Is bloodspot screening safe? What are the risks?

Screening is quick and safe. The heel prick may cause brief discomfort to your newborn, but holding or feeding them during collection will help. There is a very small risk of infection because we are making a small break in the skin, but using gloves and cleaning the heel beforehand will minimise this risk.

What are the risks of not screening? Are there any alternatives to bloodspot screening?

Affected babies who are not identified through screening will at some stage develop symptoms and could even die suddenly. While they could be offered treatment when they show symptoms, their growth and development could already be affected. Unfortunately, there are no alternatives to bloodspot screening – it is the only way to identify sick babies early.

What happens if I choose not to participate in screening?

After discussion with your midwife, if you choose not to participate in screening you will be asked to sign a 'decline of screening' form and no sample will be collected from your baby. It is recommended, for future reference, that you let your family doctor or maternal and child health nurse know about this decision.

I want my baby screened but I don't want the card used for research.

Having the screening card available for research is a personal choice and should not deter you from screening. Simply tick 'no' for the research option on the consent form.

More information

For more information you can speak to your midwife, a VCGS genetic counsellor or visit our website.

VCGS
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