What conditions does the screening detect?

There are 25 conditions covered by screening, the most commonly detected are:

- **Congenital Hypothyroidism** which, caused by a lack of thyroid hormone, can lead to poor growth and intellectual disability. If found early and treated with thyroxine tablets, the child will grow and develop normally.
- **Galactosaemia** which occurs when a baby cannot break down the sugar component of milk, known as galactose. It can cause life-threatening damage to the brain and liver within a week of birth. A special milk-free diet can prevent these problems.
- **Cystic Fibrosis** which is caused by a defective gene that results in sticky mucus in the lungs and gut. There are approved treatments that can help to prevent poor growth, chest infections and shortened lifespan.
- **Amino Acid Disorders** (such as phenylketonuria) are caused by the baby's inability to break down amino acids. Treatment with special diets and supplements can help prevent intellectual disability, seizures, organ damage and life-threatening complications.
- **Fatty Acid Oxidation Disorders** are caused when a baby is unable to convert fat into energy. Treatment with a low-fat diet, dietary supplements, and the avoidance of fasting can prevent low blood sugar and life-threatening complications.
- **Organic Acid Disorders** are caused when a baby is unable to convert amino acids into energy. Treatment with a low-protein diet and supplements can prevent vomiting, seizures and life-threatening complications.

More conditions may be added to the WA Newborn Bloodspot Screening Program in the future.

Get further information on newborn bloodspot screening from:

- Healthy WA website
- Your doctor or midwife
- WA Newborn Bloodspot Screening Program
  PathWest Laboratory Medicine WA
  PP Block, QE II Medical Centre
  Verdun Street
  NEDLANDS WA 6009
  Telephone: (08) 6383 4171
  Email: wanbs@health.wa.gov.au

For a translated brochure visit Healthy WA website

To order more copies of this brochure visit www.health.wa.gov.au/ordering

WA Newborn Screening Program

HealthyWA
healthywa.wa.gov.au

This document can be made available in alternative formats on request for a person with disability.

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Government of **Western Australia**
Department of **Health**

WA Newborn Bloodspot Screening Program

Your newborn baby's screening test

All babies are checked at birth to see that all is well and newborn bloodspot screening is part of these routine health checks

healthywa.wa.gov.au

Why is Newborn Bloodspot Screening so important?

Bloodspot screening – often referred to as the "Guthrie" or "heel-prick" test – is an important health check for your baby that can help detect serious genetic conditions that may not be obvious at birth.

The test can detect conditions in your baby **before** he or she becomes ill and while there is still time for treatment to make a difference.

About one in 1000 babies will be born with one of these conditions but most will seem healthy, showing no early signs of the underlying illness. Without early treatment these conditions can cause irreversible physical and/or intellectual disability – even death.

You do not need to have a family history of these conditions for your baby to be at risk – most babies with these conditions come from families with no history of the condition.

The screening test is strongly recommended for all newborns. Your doctor or midwife will seek your consent to perform the test and can answer any further questions you may have about the program.

The test is provided free to all babies and has been a routine part of Australian newborn care for more than 50 years. It currently finds about 35 babies with a condition in WA each year.

What does the test involve?

The test is a simple procedure usually performed when your baby is between 48 to 72 hours old. A midwife or nurse collects the blood from your baby's heel, enabling a few drops of blood to be captured on a card of blotting paper. Once dry, the card is sent for analysis at the State's pathology service, PathWest.

Should you have a home birth or leave hospital early, you will need to make arrangements with your midwife for your baby to get tested.

Why might my baby need a repeat test?

The need for a repeat test is usually due to a problem with the collection of the first sample or because the test did not give a clear result.

A request for a repeat test does not necessarily mean your baby has a condition (most needing a repeat test do not have a condition) but it is important you arrange for the repeat test as quickly as possible.

When will I get the results?

If the test results are normal, you will **not** be notified of the results, but they will be posted to your midwife or the hospital where you gave birth.

If the test shows an abnormal result, you will be contacted immediately, and you and your baby referred to a specialist. The specialist will discuss the results with you and arrange for diagnostic testing.

Does an abnormal screen mean my baby has a condition?

An abnormal result is not confirmation that your child has a condition. The bloodspot test is a screening test. As such, it identifies babies who are at increased risk of a condition.

Diagnostic testing and an examination by a specialist will be needed to determine whether your baby has that condition. This further testing needs to be performed as early as possible so if treatment is needed, it too can begin as soon as possible.

What happens to the bloodspot cards?

After testing, bloodspot cards are stored securely at PathWest's Nedlands premises for two years before being destroyed. On request in writing to PathWest, you may seek the return of your baby's card.

While in storage, the card may be used to recheck your baby's results or perform additional tests should your baby become ill. It may also be used to improve WA's screening program or develop new tests. In these instances, your baby's personal information would be removed first.

The card cannot be used in any other way without written consent from you, your baby's guardian or a lawful authority such as a court. Commonwealth Privacy Legislation and hospital and PathWest policies protect the confidentiality of all information regarding babies and their test results.

Limitations of screening

Quality assurance mechanisms ensure that through WA's Newborn Bloodspot Screening Program, bloodspot testing is available to all babies born in Western Australia and that results are valid.

Newborn bloodspot screening has been shown to be reliable but as with any laboratory test, false positive and false negative results can occur. For this reason, screening alone should never be used to rule out the possibility of a child having a condition.

If you have any suspicion your baby may have a health condition, you should follow up immediately.

The screening test for cystic fibrosis, for example, will detect only 95 per cent of babies with the condition. The test may also detect a small number of healthy babies who carry the gene for cystic fibrosis.