What conditions does the screening detect?

There are over 40 conditions covered by bloodspot screening including:

- metabolic diseases of amino acid, fatty acid, organic acid and galactose metabolism
- endocrine diseases, such as congenital hypothyroidism and adrenal hyperplasia
- cystic fibrosis
- severe combined immunodeficiency
- spinal muscular atrophy.

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

For more information about the conditions screened, scan the QR code

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, QEII Medical Centre
  Verdun Street
  NEDLANDS WA 6009
  Telephone: (08) 6383 4171
  Email: wanbs@health.wa.gov.au

For a translated brochure visit the Healthy WA website

To order more copies of this brochure visit doh.getquickmail.com

WA Newborn Screening Program

HealthyWA
Health information for Western Australians

This document can be made available in alternative formats on request.

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health.wa.gov.au

Department of Health
GOVERNMENT OF WESTERN AUSTRALIA

WA Newborn Bloodspot Screening Program

Your newborn baby's bloodspot screening test

Newborn bloodspot screening is offered to all babies. It aims to improve the health of babies by allowing early identification of those with serious but treatable conditions.

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 they become ill and while there is still time for treatment to make a positive difference.

About 1 in 600 babies are 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 increased 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. The program currently detects about 50 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, but may be taken after 24 hours. A midwife or nurse collects a few drops of blood by pricking from your baby's heel, and captures this 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 at the appropriate time.

Why might my baby need a repeat test?

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

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 babies needing a repeat test do not have a condition) but it is important your baby has this 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. 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 to confirm the screening result.

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 is 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 and screening results?

After testing, bloodspot cards are stored securely at PathWest's Nedlands premises for 2 years before being destroyed. On request in writing to PathWest, a parent or guardian may seek the return of your baby's card. Your baby's screening results will be kept securely in line with national pathology accreditation standards and may be shared with a doctor if requested and clinically required.

While in storage, the card may be used to recheck your baby's results or perform additional tests recommended by your doctor. It may also be used to help improve the WA Newborn Bloodspot Screening Program or develop new tests for conditions already approved for screening in Australia. If the results could affect the health of your baby, you will be contacted.

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

Limitations of screening

Newborn bloodspot screening has been shown to be highly 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.