Before the newborn screening test

This pamphlet is essential reading for all parents before your baby has the newborn screening test.
The pamphlet should be provided to you prior to collection of the sample to allow discussion and to obtain your consent for collection and testing.

The newborn screening test is highly recommended for all babies; however, if you choose not to have your baby tested, you should advise your doctor.

What happens to the sample cards after testing?

The newborn screening laboratory is located at Princess Margaret Hospital for Children. When testing is completed, the cards are stored securely for two years and then destroyed. The cards are sometimes used to help develop or improve new screening tests. If this happens, your baby's personal information will be removed prior to use.

The confidentiality of all information regarding babies and their test results is protected by Commonwealth Privacy Legislation and Hospital policies.

The limitations of screening

The Screening Program has quality assurance mechanisms in place to assure that all infants are screened and that the results are valid. While newborn screening has been proved reliable, as with any laboratory tests, false positive and false negative results are possible. For this reason, the possibility of a disorder should never be ruled out solely on the basis of the screening results. Any signs or symptoms of a disorder should be followed up immediately.
The screening test for cystic fibrosis will detect only 95% of babies with the disorder. The test may also detect a small number of healthy babies who are carriers of the gene for cystic fibrosis.

For more information about the screening program and the blood tests please contact:

Your doctor/midwife or

WA Newborn Screening Program

Department of Clinical Biochemistry
Princess Margaret Hospital for Children
GPO Box D184
PERTH WA 6840

Telephone: 9340 8574
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www.pmh.health.wa.gov.au/services/newborn

WA Newborn Screening Program

To order more copies of the **Your Newborn Baby's Screening Test** brochure please go to the online publication order system at www.health.wa.gov.au/ordering

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

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WA Newborn Screening Program

Your newborn baby's screening test

A blood test to screen for genetic conditions

health.wa.gov.au

Frequently asked questions

What is the newborn screening test?

This test is often called the "Guthrie test". It tests babies for serious disorders and is usually done when your baby is between 48 and 72 hours old. In Australia, the "Guthrie test" has been part of the routine care for all newborn babies for over 40 years. The testing is provided free of charge.

Why is the test done?

The test is done to find out if your baby has a disease or condition for which early treatment can prevent mental retardation, physical disability, or death. About one in every thousand babies born in Australia will have one of these treatable disorders.

How is the test done?

A midwife or nurse will perform the test by pricking your baby's heel and putting a few drops of blood on a special filter paper.

Some babies cry when their heel is pricked but the discomfort lasts only a short time. The filter paper is allowed to dry and is then sent to the newborn screening laboratory where several different tests will be performed. Make sure that your baby has this important test. If you have a home birth, check with your midwife.

What if we have no family history of the disorders?

Parents who have no family history of these problems or who have already had healthy children can still have children with these disorders. In fact, most children with these disorders come from families with no previous history of the condition.

But my baby looks healthy…

Most babies with these disorders look and act normally and seem healthy at first. The screening test helps your doctor find a problem with your baby before it makes him or her sick. Most babies who are diagnosed and treated early do well.

Why is a repeat test sometimes needed?

A few babies will need to have a repeat screening test. This is usually because there was a problem with the first sample or its collection or the test did not give a clear result. A request for a repeat test does not necessarily mean your child has a disorder. However, if you are asked for a repeat test, it is important that you take your baby for the test as soon as possible.

How will I know the results of my baby's test?

Parents are usually told of the test results only if there is a problem. If the results of your baby's test are normal, they will be mailed to the centre where your baby was born, or your midwife, about two weeks after the test. If your child's test shows an abnormal result, you will be told immediately and given instructions about what to do next.

Does an abnormal screen mean that my baby has a disorder?

Not always. Because this is a "screening test", it finds babies at increased risk for a disorder.

Your baby will need to have further testing and examination by a specialist to find out if he or she has the disorder.
The extra testing is important because early diagnosis and treatment can prevent many health-related problems.

What disorders can be detected by the newborn screen?

Phenylketonuria: caused when a baby is unable to break down the amino acid phenylalanine, which is found in the protein of foods. If detected early and the baby is started on a special low-phenylalanine diet, mental retardation is prevented.

Congenital Hypothyroidism: caused by the lack of thyroid hormone, which can lead to poor growth and mental retardation. If found early and treated with thyroid medication, the child will grow and develop normally.

Galactosaemia: occurs when a baby cannot break down the galactose part of milk sugar. In some cases, life-threatening damage to the brain and liver can occur as early as one week after birth. When started early, a special milk-free diet prevents these problems.

Cystic Fibrosis: caused by sticky secretions in the lung and gut. It is treated with dietary supplements, antibiotics and physiotherapy to help prevent poor growth, chest infections and shortened lifespan.

Amino Acid Disorders: these are caused when a baby is unable to break down certain amino acids in the blood. Treatment with special diets and supplements can help prevent mental retardation, seizures, organ damage and death.

Fatty Acid Oxidation Disorders: these are caused when a baby is unable to convert fat into energy. Treatment with a low-fat diet, dietary supplements, and avoidance of fasting can help prevent low blood sugar, coma and death.

Organic Acid Disorders: these are caused when a baby is unable to convert amino acids into energy. Treatment with a low-protein diet and supplements can help prevent vomiting, seizures, coma and death.