

## What causes these conditions?

Cystic fibrosis, PKU and other amino acid metabolism disorders, galactosaemia and FAODs are genetic disorders. These conditions are inherited from two healthy carrier parents, each carrying an abnormal gene. In most cases, the cause of congenital hypothyroidism is unknown.

## Why we test

Screening is valuable because it leads to early treatment, reducing or avoiding permanent long-term health effects. Our aim is to detect infants at increased risk of any of these conditions who need more precise and specialised diagnostic testing. It is this testing that will confirm the presence or absence of these disorders.

## Privacy

The SA Neonatal Screening Centre is bound by state regulation that requires us to keep your personal information, the test results and the blood spot sample screening cards secure. If you would like further information about the protection of these cards, please contact us.

Accessing blood spot sample screening cards for research purposes is only permitted with the approval of the Women's and Children's Hospital Ethics Committee and subject to complete anonymity.

## About us

The SA Neonatal Screening Centre is located at the Women's and Children's Hospital in Adelaide. We are committed to maintaining the highest level and quality of service, with new screening tests regularly included in pilot programs to determine whether or not these new tests are beneficial to the long-term health of babies.



SA PATHOLOGY

Quality Pathology  
supporting  
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## Further Information

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For additional information  
[www.wch.sa.gov.au/neo\\_screen.html](http://www.wch.sa.gov.au/neo_screen.html)  
[www.cyh.sa.gov.au/](http://www.cyh.sa.gov.au/)

# Screening Tests for your New Baby

*“helping to ensure  
the health of your child”*

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## About the test

Screening newborn babies to identify certain diseases is an established worldwide public health practice. The Newborn Screening Test is performed using a small sample of blood collected by a midwife when the baby is about 2 days of age. From this sample, several tests are performed to detect rare, but serious health disorders, often before there is any sign that the disorder exists. Early diagnosis and appropriate treatment can greatly reduce and often prevent the effects of the disorder for the rest of the individual's life.

To collect the sample, the baby's heel is pricked, and a small piece of special filter paper is soaked with four small spots of blood and allowed to dry. The blood spot sample card is sent to the Women's and Children's Hospital for testing.

Over 30 different health problems can be detected using these blood spots including

- Cystic fibrosis (CF)
- Congenital hypothyroidism
- Phenylketonuria (PKU)

- Galactosaemia and
- several conditions affecting the breakdown of fats (fatty acid oxidation defects - FAOD) and proteins (amino acid metabolism disorders).

## HOW OFTEN ARE THESE PROBLEMS FOUND?

Individually these health problems are rare, for example, CF affects one in every 2,500 babies.

Collectively these disorders are found in 1 out of every 800 babies born in Australia

## What we screen for

### CYSTIC FIBROSIS

Children and young people with cystic fibrosis are prone to severe chest infections and have problems digesting and absorbing food, causing serious health issues. The Newborn Screening Test detects 96% of all babies with CF. Early identification has been shown to significantly improve the life of these children although it does not prevent all health issues.

### CONGENITAL HYPOTHYROIDISM

In congenital hypothyroidism, the thyroid gland does not produce enough thyroid hormone. If not identified early, this condition causes delayed development and growth which does not improve with later treatment. Early detection and treatment with thyroid hormone tablets allows these children to grow and develop normally.

### PKU AND OTHER AMINO ACID METABOLISM DISORDERS

These are a group of inherited diseases that arise from defects in the breakdown of amino acids, the building blocks of proteins. PKU is the most common of these disorders and occurs because the baby cannot process phenylalanine. In untreated babies, high levels of phenylalanine and other chemicals formed as by-products will cause brain damage. A baby with PKU is

normal at birth but if left untreated, significant brain damage occurs within a few months. Treatment with a special diet started soon after birth allows these babies to develop normally. In addition to PKU, the Newborn Screening Test also detects rarer disorders of amino acid breakdown.

### FATTY ACID OXIDATION DEFECTS (FAOD)

Babies with a FAOD cannot use their stored fat to provide energy during times of stress, causing the body to use all its blood sugar instead. This results in potentially fatal low blood sugar levels. Children with a FAOD are generally well unless they have a viral infection (such as a cold or “gastro”), or have longer than usual time periods between meals. Treatment for this group of disorders usually involves avoiding prolonged periods of fasting and may need a special diet.

### GALACTOSAEMIA

In the body, lactose (one of the main sugars in milk) is normally broken down to galactose and glucose. Babies with galactosaemia cannot break down galactose and high levels can cause major health issues such as cataracts and liver damage. On rare occasions, if not treated, galactosaemia can be fatal. A special diet can prevent most health issues.

### HEARING

Early detection of hearing problems has been shown to benefit children's learning and development. Hearing screening of all newborn babies is now routinely performed.

### RESULTS

Test results are sent to the hospital or midwife within a week. If the results are normal you will not be notified. Occasionally, a repeat specimen may be required because of poor sample collection or a slightly elevated result. Results of repeat testing will also be sent back to your doctor or midwife.

