

The Newborn Screening Test.

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

The Newborn Screening Test blood-spot sample is collected by a midwife in the hospital where your baby is born. If your baby is not born in a hospital or if you and your baby are discharged earlier, the blood-spot sample may be collected at home. To collect the blood, 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 is sent to the Women's and Children's Hospital where it is tested and the results sent back to the hospital or midwife within a week. Your doctor or midwife will contact you if there is any suggestion that your baby might have a problem. A repeat specimen may be required because of a poor sample or for a slightly elevated result.

Over 30 different health problems can be detected using these blood-spots. These include Phenylketonuria (PKU), Galactosaemia, Congenital Hypothyroidism (CHT), Cystic Fibrosis (CF) and several conditions affecting the breakdown of fats (fatty acid oxidation defects) and proteins (amino acid metabolism disorders). Using advanced technology known as Tandem Mass Spectrometry, several tests can now be performed on each blood-spot.

HOW OFTEN ARE THESE PROBLEMS FOUND?

These health problems are rare and are found in 1 out of every 800 babies born in Australia. In SA, on average, six babies are born with CHT, and about eight babies born with CF each year. One to two babies with PKU are born each year, and one baby with Galactosaemia every two years. Fatty acid oxidation defects affect about one to two babies each year.

What are newborn babies screened for?

1 CONGENITAL HYPOTHYROIDISM (CHT)

In CHT, the thyroid gland of these babies does not produce an adequate amount of thyroid hormone. If this condition is not picked up early by the Newborn Screening Test, the problem usually does not become obvious until the baby is several months old, when children may have a delay in development and growth. Early detection and treatment with thyroid hormone tablets allows these children to grow and develop normally.

2 CYSTIC FIBROSIS (CF)

CF is an inherited problem, mostly affecting the lungs (sticky mucus) and gut (digestion of food). Children and young people with CF are prone to serious chest infections and have problems digesting and absorbing food, causing serious health issues. The newborn screening test for CF detects 96% of all babies with CF. Finding the problem early has been shown to significantly improve the life of children with CF, although it does not prevent all problems.

3 PKU AND OTHER AMINO ACID METABOLISM DISORDERS

These are a group of inherited health problems related to defects in the break down of amino acids. PKU, the most common amino acid metabolism disorder is a problem where the baby cannot break down a particular amino acid (one of the building blocks of proteins) called 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 untreated significant brain damage will occur within a few months. Treatment by a special diet started soon after birth, allows these babies to develop normally.

In addition to PKU, there are other, rarer disorders of amino acid metabolism, which the Newborn Screening Test also detects.

4 GALACTOSAEMIA

Babies with Galactosaemia cannot break down a milk sugar called galactose. In the body, lactose (one of the main sugars in milk) is normally broken down to galactose and glucose. High levels of galactose can cause major health problems such as cataracts and liver damage. Without treatment, on rare occasions, it can be fatal. A special diet can prevent most of these health problems.

5 FATTY ACID OXIDATION DEFECTS (FAOD)

Defects in the breakdown of fatty acids are a group of health problems discovered in the early 1980s. Babies with a FAOD cannot use their stored fat to provide energy during a time of stress, causing the body to use all of its blood sugar instead. This results in low blood sugar levels that can be fatal if not treated. Children with a FAOD are generally well unless they have a viral infection (cold or flu) or go for longer than usual between meals, when they may develop a severe problem. Treatment for this group of disorders usually involves a special diet and avoiding prolonged periods of fasting.

Screening tests for other disorders.

The South Australian Newborn Screening Centre is committed to maintaining the highest level and quality of service. As part of this continuing service, new developments in screening tests are regularly included in pilot programmes used to determine whether or not these new tests are beneficial to the long-term health of babies.

1 PILOT PROGRAMME FOR LYSOSOMAL STORAGE DISORDERS

The SA Neonatal Screening Centre is currently conducting a pilot study for the newborn screening of Lysosomal Storage Disorders (LSD). The study will be used to determine the effectiveness of the programme in identifying babies who are at risk of having a LSD.

The LSD are a group of rare inherited health problems, which affect 1 in 7,700 babies born in Australia. Infants with a LSD will develop a range of health problems such as bone and joint problems, breathing difficulties, developmental and behavioural problems. Early detection of these disorders enables counselling of families, close monitoring of the child and an opportunity to provide appropriate medical treatment.

What causes these conditions?

Amino acid metabolism disorders (eg PKU), fatty acid oxidation defects, galactosaemia, LSD and CF are genetic disorders, inherited from two healthy parents, each carrying an abnormal gene. In most cases, the cause of congenital hypothyroidism is not known.

What is the value of newborn screening tests?

The objective of the SA Newborn Screening Programme is to detect those infants whose tests suggest a high risk of any of these conditions and therefore need more precise and specialised diagnostic testing. It is this follow-up diagnostic testing which will confirm whether there is a problem at all. Screening is valuable because it leads to early treatment, which will lessen or avoid permanent damage to the long-term health of the child.

Storage of blood-spot cards.

The National Pathology Accreditation Advisory Council of Australia recommends that the newborn screening dried blood-spot cards be stored for 50 years. Information regarding individuals tested and the test results are securely held within the Women's and Children's Hospital in accordance with Hospital policy and guidelines governed by state confidentiality laws.

Further information.

Access to the Guthrie 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.

For more information about the neonatal screening programme, please contact:

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OR CONSULT YOUR DOCTOR.

ADDITIONAL INFORMATION LOCATED ON WEB SITES:

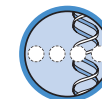
www.chempathadelaide.com/neonatal
www.cyh.sa.gov.au



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