

PILOT PROGRAMME FOR LYSOSOMAL STORAGE DISORDERS

The SA Neonatal Screening Centre is currently conducting a pilot study into the effectiveness of newborn screening for Lysosomal Storage Disorders (LSD).

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 carrier 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?

We aim to detect infants at high risk of any of these conditions who need more precise and specialised 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 reduce or avoid permanent damage to the long-term health of the child.

Storage of blood-spot cards.

The SA Neonatal Screening Centre is bound by state rules. These rules require us to keep secure information regarding individuals tested, the test results and the newborn screening dried blood spot cards. If you would like further information about the protection of newborn screening cards, please contact us.

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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ADDITIONAL INFORMATION LOCATED ON WEB SITES:

www.chempathadelaide.com/neonatal
www.chw.edu.au/prof/services/newborn/factsheets/storage.htm
www.cyh.sa.gov.au

CHILDREN, YOUTH & WOMEN'S HEALTH SERVICE



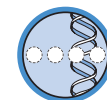
Women's
& Children's
Hospital

Department of Genetic Medicine
Women's & Children's Hospital

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Screening Tests For Your New Baby

“helping
to ensure
the health
of your child”



South Australian
Neonatal Screening Centre
Helping to ensure the health of your child

The Newborn Screening Test.

Screening newborn babies for health problems 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 either in the hospital where your baby is born or 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 for testing.

Over 30 different health problems can be detected using these blood-spots. These include Phenylketonuria, (PKU) Galactosaemia, Congenital Hypothyroidism, Cystic Fibrosis, 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.

RESULTS

The results are sent back to the hospital or midwife within a week. If all tests are normal, you will not hear of the results. A repeat specimen may be required because of a poor sample or for a slightly elevated result.

What are newborn babies screened for?

CONGENITAL HYPOTHYROIDISM

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

CYSTIC FIBROSIS (CF)

Children and young people with cystic fibrosis 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.

PKU AND OTHER AMINO ACID METABOLISM DISORDERS

These are a group of inherited health problems that come from defects in the breakdown 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.

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.

FATTY ACID OXIDATION DEFECTS (FAOD)

Defects in the breakdown of fatty acids were 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 (such as a cold or gastro) or go for longer than usual between meals. Treatment for this group of disorders usually involves avoiding prolonged periods of fasting and may need a special diet.

HEARING

Early detection of hearing problems has been shown to benefit children. Hearing screening of all newborn babies is now carried out in the newborn period. A separate pamphlet describes newborn hearing testing.

Screening tests for other disorders

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