NEWBORN SCREENING FOR GENETIC CONDITIONS

FACT SHEET 20

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Important points

- Newborn Screening (NBS) is a public health funded system for testing newborn babies' blood for about 30 rare conditions. Some of these conditions can result in physical and/or intellectual problems if not treated promptly, and are often referred to as *inborn errors of metabolism*
- The conditions tested for include phenylketonuria (PKU), congenital hypothyroidism, cystic fibrosis (CF), galactosaemia and a number of other extremely rare conditions
- This form of testing is known as *screening* because it involves testing a whole population - in this case, newborn babies. All babies are tested even if they do not have any obvious signs of a condition
- Over 99% of parents agree for their baby to have the NBS test and overall about 1 out of every 1000 babies tested (0.1%) will be diagnosed with a condition as a result of having NBS
- A few days after the baby is born, a blood sample is taken from a tiny prick of the baby's heel, placed onto a newborn screening card and the card sent to the laboratory
- A series of tests are performed on the dried blood spots. For about 1% of babies' samples, a test looks at the genetic information (the DNA) in the blood spot to clarify the result
- The parents are not notified if the test result is normal
- Each year newborn screening finds about 90 NSW/ACT babies who have one of these conditions and most of these babies benefit from treatment
- After the dried blood has been tested, it will be stored in the laboratory for varying periods in different states of Australia and in New Zealand to allow for normal quality control practices, and may need to be used for approved research after identifying information has been removed
  - The dried blood spot may be used for further testing at the request of the parent or guardian, to provide new medical information of benefit to the family
  - No further tests will be carried out on any identified stored blood sample by the newborn screening laboratory without written consent from either the parent(s)/guardian(s), the individual if they are old enough, or other lawful authority

What is newborn screening?

Newborn screening refers to the process where babies are given a simple blood test a few days after birth to see if they have a rare genetic or metabolic condition. The conditions screened for in Newborn Screening (NBS) may be life threatening and/or cause intellectual disability.

These conditions are often referred to as 'inborn errors of metabolism'.

- Metabolism is the chemical process by which food is broken down to make energy available for the normal functioning, growth and development of the body
- Enzymes are proteins that are used by the cells to break down food into a form that can be used
- Errors in metabolism occur when the essential enzymes are absent or malfunction

The aim of NBS is to detect the conditions before the onset of symptoms so treatment can be started early to reduce the effect of the condition.

This form of testing is known as *screening* because it involves testing a whole population - in this case, newborn babies. All babies are tested even if they do not have any obvious signs of a condition that affects their metabolism.

Newborn screening in Australia and New Zealand

All newborn babies are offered screening in Australia and New Zealand (Australasia).

The NBS policy for the Australasian program is developed by a joint subcommittee of the Human Genetics Society of Australasia (HGSA) and the Division of Paediatrics of the Royal Australasian College of Physicians. Newborn screening is recommended provided that:

1. There is benefit for the individual from early diagnosis
2. The benefit is reasonably balanced against financial and other costs
3. There is a reliable test suitable for newborn screening
4. There is a satisfactory system in operation to deal with diagnostic testing, counselling, treatment and follow-up of patients identified by the test

NBS to detect rare metabolic conditions is an accepted part of neonatal health care in all developed countries. NBS has been established in Australasia since the late 1960s. All Australasian newborn screening programs are fully publicly funded.

- Over 99% of parents agree for their baby to have the newborn screening test
- Overall about 1 out of every 1000 babies tested (0.1%) will be diagnosed with a condition as a result of newborn screening
- Newborn screening is a test provided for all babies free of charge

NBS services in Australasia are provided by five centralised screening laboratories. These are:

- Western Australia
- South Australia (also covers Tasmania and part of the Northern Territory)
- Victoria
- New South Wales (also covers the Australian Capital Territory)
- Queensland (also covers part of the Northern Territory)

How is newborn screening done?

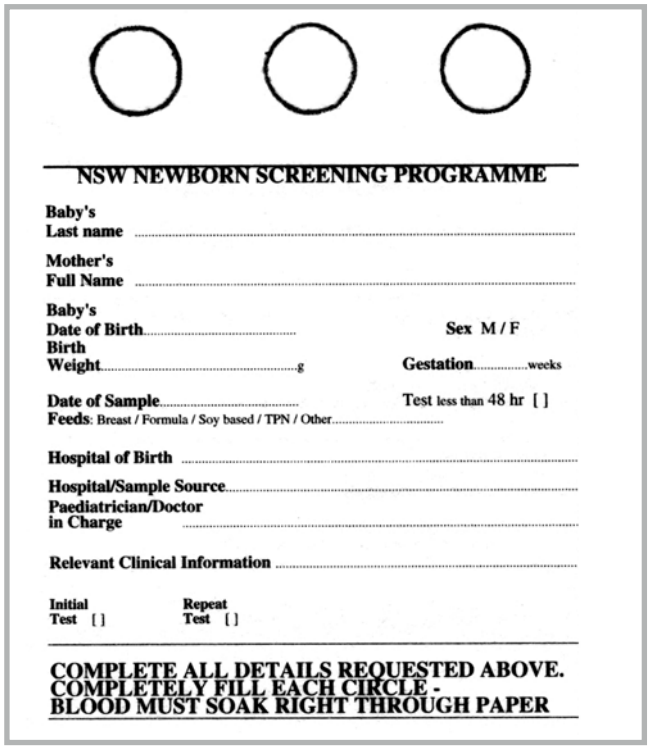
Parents are provided with information and asked for their verbal consent for the blood sample to be taken from the baby by pricking the baby's heel before the baby leaves hospital. This occurs between 48 and 72 hours after birth. For home births, the test is done on about day 4.

The small amount of blood is placed onto three different spots on a card like the one shown in *Figure 20.1* and is sent to a special laboratory.

A series of tests is performed on the dried blood spots. For about 1% of babies' samples, a test looks at the genetic information (the DNA) in the blood spot to clarify the result.

What conditions are tested for in newborn screening?

Over 30 rare conditions can now be detected by the newborn screening test. Such early detection enables treatment to be undertaken as soon as possible if a baby is shown to have the condition for which testing is done.


NSW NEWBORN SCREENING PROGRAMME

Baby's Last name .....

Mother's Full Name .....

Baby's Date of Birth..... Sex M / F

Birth Weight.....g Gestation.....weeks

Date of Sample..... Test less than 48 hr [ ]

Feeds: Breast / Formula / Soy based / TPN / Other.....

Hospital of Birth .....

Hospital/Sample Source.....

Paediatrician/Doctor in Charge .....

Relevant Clinical Information .....

Initial Test [ ] Repeat Test [ ]

COMPLETE ALL DETAILS REQUESTED ABOVE.
COMPLETELY FILL EACH CIRCLE -
BLOOD MUST SOAK RIGHT THROUGH PAPER

Figure 20.1: *The card for the newborn screening test sample (sometimes called the 'Guthrie Card') that is used for newborn screening in NSW. Small spots of blood are put onto the three circles on the card, dried and then sent to the laboratory for testing.*

For example, about 90 babies with rare medical conditions are detected each year by newborn screening in New South Wales (NSW) and the Australian Capital Territory (ACT) in Australia.

Some of the conditions for which screening is available in Australasia are

1. Phenylketonuria (PKU)

Phenylketonuria (PKU) affects about 10 babies each year in NSW/ACT

- People with PKU have a deficiency in an enzyme, produced by the liver, which usually breaks down phenylalanine, a component of protein
- The enzyme deficiency results in a build-up of phenylalanine in the blood and can cause severe brain damage if untreated
- Detection in newborn babies allows early treatment with a special milk formula and a special diet when the baby is older so that the baby will have normal growth and development

The newborn screening for PKU is illustrated by Jenny's story:

Baby Nicola looked healthy when she was born, so Jenny was shocked to hear her baby was at risk of growing up with brain damage if she did not have a special diet. Jenny knew nothing about PKU but it is one of the most important diseases that newborn screening detects because treatment makes such a difference to the lives of these babies as they grow up. Now Jenny has seen the doctors and other specialists, she feels she can cope with the special diet Nicola will need to follow to protect her brain from permanent damage. On the special PKU diet, Nicola's brain will develop normally and she will have the chance to do well at school like other children.

2. Congenital hypothyroidism

Congenital hypothyroidism affects about 26 babies each year in NSW/ACT.

- It is caused when the thyroid gland is either too small, absent or does not work properly
- Early treatment leads to the normal mental and physical development of the child. Treatment is a small tablet of thyroid hormone given daily

3. Cystic fibrosis (CF)

Cystic fibrosis (CF) affects about 34 babies each year in NSW/ACT.

- In people with CF, their bodies produce thicker mucus than normal in the bowel and the lungs
- This can cause chest infections or diarrhoea and could stop the baby gaining weight
- Recent medical and scientific advances have greatly improved the outlook for these babies (see Genetics Fact Sheet 33)

4. Galactosaemia

Galactosaemia affects about 1-3 babies each year in NSW/ACT.

- In people with galactosaemia, one of the sugars (galactose) contained in milk (both breast and cow's milk) accumulate in the blood

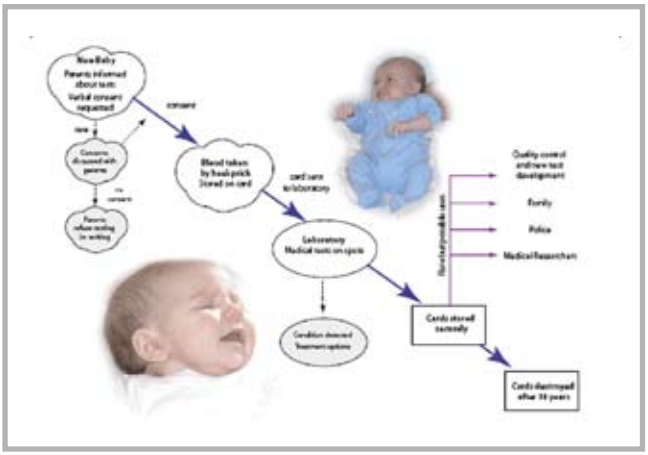


Figure 20.2: A summary of the newborn screening process

- Serious illness will be prevented if the child is treated promptly with special milk, which does not contain galactose
- Babies with this condition, if not treated, may become very sick and die

5. Other conditions

New technology has made it possible to detect a number of extremely rare conditions, using the dried blood sample, so that treatment can be started early.

These very rare medical conditions affect overall about 15 babies each year.

- They include a range of conditions due to problems with important components of food such as fatty acids, organic acids and amino acids defects
- Early detection is important as diet and medications can treat most of these conditions that, without appropriate treatment, can cause severe disability or death
- The most common of these conditions is MCAD (medium-chain acyl CoA dehydrogenase deficiency)

What happens if the blood test shows that a baby is affected with one of these conditions?

Parents are not notified if the test result is normal.

About 1 baby in every 100 will need a second blood test if the first test did not give a clear result. Parents will be notified if a second test is needed. The second test will almost always give a normal result, and the doctor will be sent the result.

In a small number of cases the blood test will be abnormal, further investigations will be necessary, and treatment may be needed. The family doctor will be told about the result and generally organise testing or referral (see *Figure 20.2*).

Contact the local genetic counselling centre for more information regarding available testing in your state or country and the risks for future children where there are affected family members (see Genetics Fact Sheet 3).

What happens to the test sample?

After the dried blood has been tested, it will be stored in the laboratory. No further tests will be performed on any identified stored blood sample by the newborn screening laboratory unless there is written consent from either the parent(s)/guardian(s), the individual if they are old enough, or other lawful authority.

The stored samples will be retained to allow for normal quality control practices, and may need to be used for approved research after identifying information has been removed. It may be used for further testing at the request of the parent or guardian, to provide new medical information of benefit to the family.

Other Genetics Fact Sheets referred to in this Fact Sheet: 3, 33

Information in this Fact Sheet is sourced from:

Joint newborn screening committee of the Human Genetics Society of Australasia (HGSA) and Division of Paediatrics of the Royal Australasian College of Physicians(2004) [online]. Available from: http://www.hgsa.com.au/ [Accessed June 2007]

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