

Test results

All of the disorders listed in this brochure are rare, and in most cases the test results are normal. Parents are contacted if there are concerns with the test results. The receipt of each baby's sample is confirmed with the hospital.

A few babies will need to have a second blood test. This is usually because the first test did not give a clear result. Most babies' second tests will give normal results and your doctor will be informed.

In a very small number of babies the blood test will be abnormal. Parents will be notified by their doctor or midwife if there is a need for further tests, and if necessary, treatment of your baby. Ask your baby's doctor to give you the test information fact sheet sent to them by the laboratory.

Further information can also be found on the NSW Newborn Screening Programme website www.chw.edu.au/prof/services/newborn or by telephoning (02) 9845 3659.

Screening tests for other disorders (Pilot Programs)

The NSW Newborn Screening Programme is committed to maintaining the highest possible standards. To improve the quality of its service, new developments in other parts of the world are regularly investigated in pilot programs, to find out whether or not the new tests would be beneficial to babies born in NSW or ACT.

As well as the screening for above congenital disorders, your baby will also be offered hearing screening.

PRIVACY NOTICE

The NSW Newborn Screening Programme complies with the *Privacy and Personal Information Protection Act* including requirements for secure storage of information and limited disclosure. Parents have the right to access information associated with the screening process. Parents may refuse the newborn screening test on behalf of their baby and will be required to sign the hospital's disclaimer form. However, the programme diagnoses about ninety babies each year and refusal of the test might unnecessarily risk the baby's health if urgent treatment is needed.

You are encouraged to take your baby to your local Early Childhood Health Centre for regular check-ups and immunisation

Look under 'E' for Early Childhood Health in the *White Pages* of the phone book.

FOR FURTHER COPIES PLEASE CONTACT:

Better Health Centre – Publications Warehouse
Locked Mail Bag 5003 Gladesville NSW 2111
Tel. (02) 9816 0452 Fax. (02) 9816 0492
TTY. (02) 9391 9900

A full copy of this report can be downloaded from the NSW Health website. www.health.nsw.gov.au



TESTS TO PROTECT YOUR BABY



Why early diagnosis
is important to
your baby's future

IMPORTANT

Please keep this information for three months after your baby is born. You may receive a request to have the test repeated or your doctor may need to follow up your baby's test results with further investigations.

Early diagnosis

Some rare disorders in an apparently healthy baby can be detected by a blood test done between 48 to 72 hours after birth. Early diagnosis means that treatment can be started quickly, before the baby becomes sick.

In NSW and the ACT, this blood test is part of the normal care of newborn babies, and in fact is an established practice worldwide.

The testing is provided free-of-charge, and is funded by the NSW and ACT Departments of Health. Shortly before each baby leaves hospital, a few drops of blood are taken from the baby's heel and collected onto special absorbent paper. The dried blood sample is sent to the NSW Newborn Screening Laboratory at The Children's Hospital at Westmead for testing. Over thirty different congenital disorders can now be detected using the baby's blood sample, and these are described in this leaflet.

Make sure that your baby has had this important test. If you have a home-birth, check with your midwife.

Make sure it is noted that the test has been carried out in the 'birth details' section of the baby's *Personal Health Record*.

The National Pathology Accreditation Advisory Council (NPAAC) requires newborn screening samples to be stored. After the dried blood spot has been tested, it will be stored in a secure locked area until the child is 18 years of age,

when the sample will be destroyed. The stored sample is retained to allow for normal quality control practices, and may be used for ethics committee approved research after identifying information has been removed. The sample may also be used for further testing at the request of the parent or guardian, to provide new medical information for the benefit of the family. However, the NSW Newborn Screening Laboratory will not perform any further tests on any identified stored blood sample without written consent from the parents/guardians or other lawful authority in exceptional cases such as if ordered by a court.

What are the disorders?

CONGENITAL HYPOTHYROIDISM

Hypothyroidism affects about one in 3,500 babies. It is caused by an absent, small or improperly functioning thyroid gland. Lack of thyroid hormone slows brain development and growth. Early treatment with daily thyroid hormone leads to normal mental and physical development.

PHENYLKETONURIA (PKU)

PKU is a rare condition which affects about one in 10,000 babies. A baby with this problem cannot properly use one of the building blocks of protein called phenylalanine, which accumulates in the blood and causes brain damage. If the baby is given a diet low in phenylalanine (very low protein plus a special formula) normal development will occur.

OTHER DISORDERS OF PROTEIN AND FAT METABOLISM

Tandem Mass Spectrometry, introduced in 1998, detects a number of extremely rare disorders using the dried blood sample. These are disorders of the metabolism of protein and fat. About one in 6,000 babies is found with one of these rare disorders. If detected early they can be treated, usually by modification of the baby's diet, and avoiding periods of fasting. If your baby requires a repeat test more information

will be given about the specific disorder which is suspected. Without appropriate management, some disorders can cause severe disability or death.

CYSTIC FIBROSIS (CF)

One in every 2,500 babies has cystic fibrosis. In cystic fibrosis, the mucus produced in the intestines and lungs is thicker than normal. This results in infections in the lungs. In the intestines, it may lead to difficulties with digesting food properly. Babies with CF may have a cough when very young and may be hungry but gain weight poorly.

The newborn screening test detects about 95 percent of babies with CF but also detects a few babies who are HEALTHY carriers of CF. Babies with a positive test result will need a sweat test at about six weeks of age to determine whether the baby has CF or is only a healthy carrier. A healthy carrier is not affected with CF and their body functions normally.

Recent medical and scientific advances have greatly improved the outlook for babies with cystic fibrosis. Early diagnosis and treatment are important.

GALACTOSAEMIA

Galactosaemia is an extremely rare disorder only affecting one in 40,000 babies. The disorder is caused by the accumulation of galactose (a type of sugar in milk) in the blood. Prompt treatment with a special milk, which does not contain galactose, will completely prevent serious illness. Untreated babies with galactosaemia may become very sick and die.



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