

**CIRCULAR**

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**Guidelines for Newborn Screening**

This Circular supersedes Circular 2001/45, also referenced in the Patient Matters Manual Chapter 2, Paediatrics, point 2.1, page 2.1.

The Circular contains revised information about numbers of disorders detected; use, storage and retention of samples; and, hospital protocols concerning provision of information, obtaining consent and collection of the sample.

Hospitals with maternity units should ensure that newborn screening tests are offered to all newborn babies. Newborn screening can detect a number of serious genetic disorders such as phenylketonuria, primary congenital hypothyroidism, cystic fibrosis, galactosaemia and more than 30 rarer metabolic disorders, involving fatty acid, amino acid and organic acid defects. Among the 87,000 babies born each year in NSW, about 90 are diagnosed with one of these genetic conditions.

Early diagnosis and immediate treatment by medication or diet can prevent death or serious illness including mental retardation, and can lead to significantly improved outcomes.

The test is carried out on a blood sample, which is obtained by heel prick, placed on special pre-printed filter paper and processed at the NSW Newborn Screening Laboratory.

Before the newborn screening test is carried out, staff must ensure that parents or guardians are properly informed about the test and its importance and that consent is obtained.

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Distributed in accordance with circular list(s):

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In accordance with the provisions incorporated in the Accounts and Audit Determination, the Board of Directors, Chief Executive Officers and their equivalents, within a public health organisation, shall be held responsible for ensuring the observance of Departmental policy (including circulars and procedure manuals) as issued by the Minister and the Director-General of the Department of Health.

Parents or guardians should also be advised that the National Pathology Accreditation Advisory Council (NPAAC) require newborn screening samples to be stored. After the dried blood spot has been tested, it will be stored by the laboratory in a secure locked area until the child is 18 years of age, when the sample will be destroyed. However, no further tests will be performed on any identified stored blood sample by the newborn screening laboratory without written consent from the parents/guardians, the child, if old enough, or other lawful authority. Parents have the right to access information associated with the screening process, and the NSW Newborn Screening Programme complies with the Privacy and Personal Information Protection Act.

**All hospitals with maternity units are required to develop written policy and procedures, incorporating the information in this circular, to ensure that newborn screening is offered to all infants. The information that follows is provided to assist hospitals in developing these protocols.**

Robyn Kruk  
**Director-General**

# Guidelines for Newborn Screening

## 1. Conditions detected by newborn screening

Newborn screening tests should be offered to all newborn babies in New South Wales to ensure the early detection and treatment of phenylketonuria, primary congenital hypothyroidism, cystic fibrosis, galactosaemia and more than 30 rarer metabolic disorders, involving fatty acid, amino acid and organic acid defects.

**Phenylketonuria** occurs in about 1 in 10,000 live births (about 10 babies per year in NSW). If the baby is given a diet low in phenylalanine, normal development will occur. Without treatment, severe brain damage results.

**Primary congenital hypothyroidism** occurs in about 1 in 3,500 live births (about 26 babies per year in NSW).

For both these conditions, delays in starting treatment are associated with a poorer intellectual outcome. The aim is to have all babies on treatment as soon as possible and preferably by 10 days of age.

**Cystic fibrosis** affects about 1 in 2,500 live births (about 34 babies per year in NSW). The aim is to start treatment by at least 2 months of age.

**Galactosaemia** occurs in about 1 in 40,000 births (about 1-3 cases per year in NSW). Immediate treatment may be life saving.

**Some fatty acid, organic acid and other amino acid defects** can now be detected using Tandem Mass Spectrometry. These much rarer metabolic disorders affect about 15 babies per year. Early detection is important as diet and medications can treat most of these disorders. Without appropriate management they can cause severe disability or death.

## 2. Informing parents and guardians about the proposed tests

Before the newborn screening test is carried out, **staff must inform parents or guardians about the test and its importance**. This information is included in the pamphlet "Tests to protect your baby" which staff must discuss with parents or guardians either in a one to one or group situation. **Distribution of the pamphlets without discussion is not permissible**. The pamphlets are available from:

The Better Health Centre – Publications Warehouse  
Locked Mail Bag 5003  
GLADESVILLE NSW 2111  
Telephone: (02) 9816 0452  
Facsimile: (02) 9816 0492

**Two points** must be made clear, prior to the sample collection.

- **Storage** – After testing, the blood spots are stored by the laboratory in a secured locked area. No further tests will be performed on any identified stored blood sample by the Newborn Screening Laboratory without written consent from both parents / guardians. The stored sample will be retained to allow for normal quality control practices, and may be used for approved research after identifying information has been removed. It may also be used for further testing at the request of the parents or guardians, to provide new medical information for the benefit of the family. The National Pathology Accreditation Advisory Council (NPAAC) requires samples to be stored. The NSW Newborn Screening Programme will store samples until the child is 18 years old.
- **Privacy** – The laboratory complies with the NSW Department of Health’s Information Privacy Code of Practice and the Privacy and Personal Information Protection Act including the requirements of storage of information and limited disclosure. Parents have the right to access information associated with the procedure.

### 3. Consent

As with all medical procedures or treatments, the test should comply with “Patient Information and Consent to Medical Treatment” – NSW Health Department Circular 99/16.

Each hospital’s newborn screening policy and procedures must include instruction on gaining verbal consent from parents or guardians. There must be documentation on the mother’s file stating there has been discussion about the newborn screening test. The file should also show a record of completion of the newborn screening test. A pre-inked stamp similar to the example below is recommended. All parts need to be completed.

<b>Baby’s name:</b>	
<b>Signature (Health Professional)</b>	
1. <b>Discussing NBS information:</b> _____	<b>Date:</b> _____
2. <b>Provision of the NBS pamphlet:</b> _____	<b>Date:</b> _____
3. <b>Completion of NBS test:</b> _____	<b>Date:</b> _____

Parents may refuse the newborn screening test on behalf of the baby. However, the program diagnoses about 90 babies each year for which treatment is urgently needed and refusal of the test might unnecessarily risk the baby’s health. Hospitals should devise a protocol for parental refusal of a newborn screening test for their baby. It should include parents being properly informed about the test and its importance, the paediatrician also talking with the parents and listening to their concerns, and an offer of the option of telephoning the Director/Head of the Newborn Screening Programme to answer any further questions they may have. Refusals must

be documented and signed in the mother's/baby's file and a newborn screening sample card with completed demographic information, and "refusal" written on it sent to the laboratory. This is important for legal protection of both the hospital and the laboratory.

#### 4. Timing of the newborn screening test

It is recommended that the newborn screening test be carried out when the baby is between 48 and 72 hours old. False positive and false negative results sometimes occur when the tests are done before 48 hours.

The period of hospitalisation provides the only certain opportunity for testing. If the baby is discharged before 48 hours, the test must be carried out before the baby leaves hospital, unless the hospital of birth has a failsafe domiciliary midwifery program (DMP). The test may then be done in the community as soon after 48 hours as possible.

***It is the responsibility of the hospital of birth to ensure that a newborn screening sample is taken by checking the confirmation report, especially when the baby is referred to a postnatal service outside the area.***

#### 5. Warming the heel

Variation in practice has been noted in the methods used in preparation for drawing blood from neonates. Some practices eg (***warming the foot with water***), are associated with foreseeable risks ***such as burns*** and therefore should be avoided.

Hospitals should review local practices to identify methods that may carry an avoidable risk and replace these with safer alternatives such as taking the sample after a bath, the use of booties, the use of a foot warmer and/or overhead heating to improve blood flow to the heel.

#### 6. Twin to twin transfusion

It is possible to miss a disorder in a twin who had twin to twin transfusion. It is essential that this information be written on the card, so that the test will be repeated.

#### 7. Conducting the test

The newborn screening test is carried out on a blood sample obtained by heel prick. The blood sample is placed on a special pre-printed filter paper card provided by the NSW Newborn Screening Programme and processed by the Programme at The Children's Hospital at Westmead.

It is essential that ***all information is completed on the newborn screening card***. Each field has been included for a specific purpose.

The laboratory issues supplementary advice to maternity units, on procedures for collecting and handling newborn screening samples. This information is available from:

The NSW Newborn Screening Programme  
Locked Bag 2012  
WENTWORTHVILLE NSW 2145  
Telephone: (02) 9845 3255 / 3659  
Facsimile: (02) 9845 3800

## 8. Test results

Test results are normally available 24 hours after the sample is received in the laboratory.

### ▪ **Urgent Follow-up**

Test results which are clearly indicative of any one of the disorders are notified by telephone to the paediatrician/doctor/independent midwife shown on the newborn screening test card.

***This name must be filled in.*** It is the responsibility of this person to ensure that the baby is promptly referred for further investigation and treatment. Where there is uncertainty regarding whose name is to be written on the card, it is recommended that the name be that of the ***paediatrician of the day.***

### ▪ **Retesting**

Retesting is carried out in the following circumstances:

- Moderately elevated results, which are likely to be of a transient nature but require confirmation (a written request for resample is sent by the laboratory).
- Insufficient/contaminated test samples (a written request for resample is sent).
- When blood is collected before 48 hours, ***it is expected the hospital will automatically send another sample*** collected between 48 – 72 hours.

## 9. Hospital nominated newborn screening liaison person

Hospitals should ensure that a nominated person (eg community liaison midwife or nurse manager of maternity) is responsible for handling telephone calls and correspondence concerning newborn screening and the organisation of retesting. This nominated person should be covered for holiday/sick and other types of leave. The name and position of the nominated person should be notified in writing to the:

The NSW Newborn Screening Programme  
Locked Bag 2012  
WENTWORTHVILLE NSW 2145  
Telephone: (02) 9845 3255 / 3659  
Facsimile: (02) 9845 3800

### **Responsibilities of the nominated newborn screening liaison person;**

- Acting as the contact for NSW Newborn Screening Programme.
- Checking the quality of blood samples and data on cards before arranging for cards to be **sent daily** by post or by courier.
- Contacting the parents when a resample is necessary and giving them the Fact sheets, which are provided by the laboratory with the request to resample.
- Informing the baby's doctor of the need and indication for the resample.
- Ensuring that the **confirmation report is checked against the hospital births as soon as is possible.**

The laboratory issues **confirmation reports** to hospitals approximately every 2 weeks. These reports contain a list of babies for whom samples have been received for the previous fortnight. The nominated person should contact the laboratory concerning any baby whose name is missing from the list and check if the sample has been received.

If a baby has not had a sample collected the nominated person is responsible for arranging a sample collection. The reason the baby was missed should be documented in the file.

**The hospital of birth is responsible for ensuring all babies are offered the newborn screening test. This includes babies who are transferred to other hospitals or domiciliary midwifery programs.**

Robyn Kruk  
**Director-General**

## References:

### 1. Tests to protect your baby

Ref no: (HPA) 980026, NSW Health Department, available from:

The Better Health Centre – Publications Warehouse  
*Locked Mail Bag 5002*  
GLADESVILLE NSW 5003  
Telephone: (02) 9816 0452  
Facsimile: (02) 9816 0492

### 2. NSW Newborn Screening Programme advise on procedures for collecting and handling newborn screening samples, available from:

The NSW Newborn Screening Programme  
Locked Bag 2012  
WENTWORTHVILLE NSW 2145  
Telephone: (02) 9845 3255 / 3659  
Facsimile: (02) 9845 3800

**NSW Newborn Screening website** <http://www.chw.edu.au/prof/services/newborn>

### 3. NSW Department of Health

The following information is available on NSW Health's website: [www.health.nsw.gov.au](http://www.health.nsw.gov.au).

- Information Privacy Code of Practice, December 1998
- NSW Department of Health Circular No 99/34 – State Records Act 1998
- NSW Department of Health Circular No 98/59 – Principles for Creation, Management, Storage and Disposal of Health Care Records.
- NSW Department of Health Circular 99/78 – General Disposal Authority 5 – Public Health Services: Patient/Client Records (GDA 5)
- Patient Information and Consent to Medical Treatment – NSW Health Department Circular 99/16.