NSW Newborn Screening Programme

Sampling Information And Guidelines

Notes for midwives, clinical nurse educators, early childhood/community health nurses, staff in pathology laboratories, and other health professionals
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INTRODUCTION

The newborn screening test is an essential part of normal newborn care.

Staff in maternity units, independent midwives, early childhood/community health nurses and doctors should ensure that all newborn babies are offered a newborn screening test.

The NSW Newborn Screening Programme screens all babies born in NSW and ACT (about 92,000 babies per year), and about 90 of these are diagnosed with a serious genetic metabolic disorder. There are Newborn Screening Programs in all states of Australia.

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

Certain rare, but treatable disorders, have few specific clinical indicators in the newborn baby. Newborn screening programs allow for early diagnosis and immediate treatment by medication or diet which can prevent serious complications such as mental retardation or death, and can lead to significant improved outcomes.

Dried blood samples collected from all newborns are tested for:-

- Phenylketonuria (PKU)
- Primary Congenital Hypothyroidism (CH)
- Cystic Fibrosis (CF)
- Galactosaemia(s)
- More than 30 fatty acid, amino acid and organic acid disorders

Any other disorder recommended by the NSW Newborn Screening Advisory committee may be incorporated into the newborn screening programme as a pilot study.

(See Attachment 1 - Specific problems that can occur with each disorder)
(See Attachment 2 - Clinical aspects of disorders)

<table>
<thead>
<tr>
<th>DISORDER</th>
<th>START DATE</th>
<th>INCIDENCE</th>
</tr>
</thead>
<tbody>
<tr>
<td>Phenylketonuria (PKU)</td>
<td>1964</td>
<td>1:10,000</td>
</tr>
<tr>
<td>Hypothyroidism (CH)</td>
<td>1977</td>
<td>1:3,500</td>
</tr>
<tr>
<td>Cystic Fibrosis (CF)</td>
<td>1981</td>
<td>1:2,500</td>
</tr>
<tr>
<td>Galactosaemia</td>
<td>1983</td>
<td>1:40,000</td>
</tr>
<tr>
<td>&gt; 30 rare metabolic disorders</td>
<td>1998</td>
<td>1:5,600</td>
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*There are about 90 babies diagnosed each year with one of these serious genetic metabolic disorders.*

Newborn screening tests are based on the analysis of biochemical markers that accumulate if the baby has one of the above disorders. The levels of these vary in the first days after birth. **The optimum time for collection is between 48 and 72 hours, so as to be sure to diagnose the disorders before the onset of any adverse effects.** False positive and negative results may occur when the tests are done before 48 hours. (attachment 1)

The period of hospitalization provides the only certain opportunity for testing. If the baby is discharged before 48 hours, the test should be carried out before the baby leaves hospital, **unless** the hospital of birth has a failsafe community midwifery program.
SPECIFIC PROBLEMS IN SCREENING FOR EACH DISORDER

- **PHENYLKETONURIA (PKU)** can be missed if the sample is taken before the baby is 48 hours old. Babies with phenylketonuria have a normal phenylalanine level at birth (accumulation of phenylalanine does not occur in utero because of the mother’s normal metabolism).

- **PRIMARY CONGENITAL HYPOTHYROIDISM**: Thyroid Stimulating Hormone (TSH) can be elevated if the sample is taken before baby is 24 hours old because there is a TSH surge on day 1. Of babies tested on day 1, 2% have falsely elevated results and require a second test.

- **CYSTIC FIBROSIS (CF)**: The screening protocol for cystic fibrosis involves a test for immunoreactive trypsin (IRT), with elevated results having a specific DNA test. This protocol will result in the identification of about 95% of CF cases. About five percent of CF cases each year will be missed (approximately 2 babies per year). The initial screening test is not valid if the sample is collected after 8 weeks of age, as the IRT level falls with age.

- **GALACTOSAEMIA** could possibly be missed by our test protocol if the baby has not had at least one milk feed before the sample is taken. Babies with classical galactosaemia have no symptoms in the first 48 hours. Milk feeding is not required before a newborn screening sample is collected regardless of weight and maturity of the baby. Information on the card must be completed especially the feeds given at the time of sampling. It must be made clear on the card whether the baby is on a cow’s-milk based or soy-based formula, for example Karicare has 2 formulae, one cow’s-milk based and one soy-based. However, if a baby has suggestive symptoms and is not on milk feeds (breast/cow-based formula), the laboratory can perform an enzyme test if notified of clinical symptoms provided the baby has not had a blood transfusion.

- **OTHER RARE METABOLIC DISORDERS**: over 30 rare metabolic disorders such as amino acid, organic acid and fatty acid oxidation defects are screened for using tandem mass spectrometry. The incidence of these three groups of disorders, excluding phenylketonuria, is 1:5,750. These disorders are autosomal recessive, resulting in an enzyme deficiency (enzymes are essential for the body’s metabolism). All these rare metabolic disorders which can be detected by newborn screening are treatable either by diet and/or medication. Fact sheets about each separate disorder are available for health professionals.
PHENYLKETONURIA (PKU) is a rare, recessively inherited inborn error of phenylalanine metabolism. Babies are normal at birth, but blood phenylalanine levels rise quickly, and central nervous system damage occurs. Without treatment, severe mental retardation is the usual outcome. A low phenylalanine diet instituted soon after birth ensures normal development. The diet is complex, and in NSW the only PKU clinic is at the Children’s Hospital at Westmead. The screening test for PKU measures the blood phenylalanine level using tandem mass spectrometry.

PRIMARY CONGENITAL HYPOTHYROIDISM (cretinism) is caused by the absence or abnormal formation or function of the thyroid gland. Without early treatment both intellectual delay and growth retardation can occur. The intellectual delay is usually not severe, but is irreversible. Early treatment with thyroid hormone results in normal growth and development. Congenital Hypothyroidism is detected by a test which measures thyroid stimulating hormone (TSH). If an elevated level of TSH is found the baby requires plasma thyroid function tests and a thyroid scan/ultrasound so that a precise diagnosis can be made. An x-ray of the knee for bone age is also useful to identify those babies (with retarded bone age at birth) who are at risk of mild developmental problems despite early treatment.

CYSTIC FIBROSIS is a recessively inherited disorder of chloride and sodium transport across cell membranes. It is characterised by chronic suppurative lung disease, and pancreatic insufficiency which causes failure to thrive. Cystic fibrosis is detected by a test which measures immunoreactive trypsin (IRT) in the blood sample. If the IRT level is elevated the same sample is analysed for the most common cystic fibrosis DNA mutation ΔF508. There are over 900 other CF mutations, some of which give the child a much milder form of Cystic Fibrosis. It is the babies with the milder mutations that may be missed by newborn screening (approximately 2 babies each year).

Early diagnosis enables early treatment, thus avoiding the early morbidity, which may be otherwise associated with cystic fibrosis, and permits genetic counselling and family planning.

GALACTOSAEMIA: Babies with recessively inherited galactose-1-phosphate uridyl transferase deficiency (classical galactosaemia) cannot process galactose, a component of lactose. Life threatening liver failure and infections can occur. A galactose-free diet in the first week is life saving and is continued throughout life. Galactosaemia is currently detected by a test, which measures blood levels of galactose and galactose-1-phosphate which if elevated is followed up by a conclusive enzyme test for galactose-1-phosphate uridyl transferase which will be completely deficient in this disorder.

OTHER RARE METABOLIC DISORDERS: Newborn Screening is using tandem mass spectrometry to test for over 30 rarer metabolic disorders of fatty acid, organic acid, and amino acid defects. These disorders include medium chain acyl CoA dehydrogenase deficiency (MCAD), maple syrup urine disease (MSUD), methylmalonic aciduria (MMA) and many more. They are all genetic disorders caused by an enzyme defect. These disorders are all treatable with either dietary management and/or medication. Without appropriate management these disorders can cause severe disability or death.
GUIDELINE (1)
CARE OF BLANK NEWBORN SCREENING
SAMPLE CARDS

The Newborn Screening cards are special pre-printed filter paper obtained from the:

NSW Newborn Screening Laboratory

Telephone No: (02) 9845 3255
Fax No: (02) 9845 3800
Address: NSW Newborn Screening Programme
Locked Bag 2012
WENTWORTHVILLE NSW 2145

BEFORE SAMPLE COLLECTION

- Store cards in clean, dry area.
- Store cards in the dark or normal light (not direct sunlight).
- Remove cards from area if near any type of fumes (eg: paint/varnish/glue/organic solvents – fumes may fix the haemoglobin and make testing impossible) This is very important.
- When handling cards, before or after the sample is taken, DO NOT TOUCH blood circle area. (possibility of contamination).
GUIDELINE (2)
INFORMING AND GAINING VERBAL CONSENT FROM PARENT/GUARDIAN

1. Informing parents/guardians about the proposed tests

- The pamphlet “Tests to protect your baby” [SHPN (COM) 030194 revised edition July 2003 and reprinted edition January 2005] are the only pamphlets to be handed to parents. ALL OTHER PAMPHLETS TO BE DICARDED. The pamphlet is available from the Better Health Centre.

  Telephone: (02) 9816 0452 Facsimile: (02) 9816 0492
  TTY: (02) 9391 9900 (for the hearing impaired).

- A shortened version of the pamphlet has been translated into 9 languages (Arabic, Chinese, Indonesian, Japanese, Khmer/Cambodian, Korean, Serbian, Turkish and Vietnamese) and is available from the Newborn Screening Laboratory and the website:
  Telephone: (02) 9845 3659 Facsimile: (02) 9845 3800

- The information in this pamphlet MUST BE DISCUSSED with parent on a one to one or group basis.

- The information about newborn screening should be discussed some time during the pregnancy AS WELL AS before the sample is collected.

- Information to discuss is:
  - Importance of newborn screening
  - Disorders screened
  - Test results – usually available 24-48 hours after the sample is received in the laboratory
  - Pilot programs
  - Storage of cards for 18 years in a securely locked area and then destroyed – See “Newborn Screening in New South Wales: Storage of samples”. (attachment 3)
  - Reasons for storage – normal quality control / laboratory audit / develop new tests / ethics committee approved research after de-identifying information / for family use (attachment 3)
  - No further tests other than the newborn screening tests discussed in the pamphlet will be performed without the written consent of both parents
  - Newborn Screening complies with the Privacy and Personal Information Protection ACT (1988) and parents and guardians have the right to access information associated with the screening process
  - Verbal consent

2. Gain verbal consent from parent/guardian

Verbal consent must be given by parent/guardian prior to sample collection
“Patient Information and Consent to Medical Treatment – NSW Health Department Policy Directive PD2005_566”
If parent refuses see refusal guideline (attachment 6)
NEWBORN SCREENING IN NEW SOUTH WALES: STORAGE OF SAMPLES

All babies in NSW and the ACT are offered testing for treatable disorders. Newborn screening samples of blood, dried on filter paper, are collected from all babies in New South Wales and the Australian Capital Territory, and tested for a number of metabolic disorders for which early treatment will prevent mental retardation and other serious medical problems. About 90 babies each year are diagnosed with a treatable condition. In many of these, failure to diagnose the condition in the newborn period would lead to irreversible problems, such as mental retardation or, more rarely, death.

Screening cards are stored until the child reaches 18 years of age. The dried blood samples are stored after the testing is completed, until the child reaches 18 years, at which time the cards are destroyed. The storage is explained in the newborn screening pamphlet handed to parents, “Tests to protect your baby”.

The reasons for storage are:

- **For laboratory audit.** If a baby is later found to have a disorder that was missed by the newborn screening test, the laboratory needs to know what went wrong, so as to be able to rectify the problem.

- **To develop new tests.** The screening programme must be able to develop new tests for treatable conditions. If there is a disorder normally recognised during childhood, when damage has already occurred, it is necessary to know what the newborn blood sample showed, to see if the disorders could have been diagnosed by a newborn test. This is the major reason why blood samples are stored in identifiable form.

- **For family use.** Some families are able to make use of stored samples. If a child died from an unknown disorder, sometimes a likely diagnosis emerges later. The stored sample may be able to be used to confirm the diagnosis. This helps families come to terms with what has happened, and may be useful for prenatal diagnosis.

Samples are stored securely in a locked area. Only authorised staff from the screening programme can access them. The data stored is also secured, by multiple password systems. The stored data consists of very basic demographic data – name, date of birth, hospital of birth, birth weight, baby’s doctor, plus the results of tests.

There is no stored data about DNA. About 1% of the samples has a test for a common change in the DNA (a mutation) associated with two of the disorders, as part of the routine testing. No DNA tests are done on the vast majority of samples, and absolutely no other DNA records are held.

No tests other than routine newborn screening tests are carried out on any identified sample without the written permission of the parents or guardian, or the subject, if old enough.

There is a memorandum of understanding between NSW Police and NSW Health that samples would only be sought by police to identify remains. The police have only very occasionally requested access to samples, for identification of a deceased person, and in each case, with written permission from parents. In general it is only thought useful to access samples for forensic purposes if the subject is dead, or believed dead, and when the newborn screening card is the only available sample to help in identification.

The newborn screening programme complies with the Privacy and Personal Information Act of 1998. These issues have been discussed with the Privacy Commissioner. It is considered that the present policy of storing samples is of benefit to the children of New South Wales and the Australian Capital Territory rather than a risk.

Professor Bridget Wilcken
Clinical Director, NSW Newborn Screening Programme

Dr Veronica Wiley
Head, NSW Newborn Screening Programme
GUIDELINE (3)
SAMPLE COLLECTION PROCEDURE

The Newborn Screening sample should be taken between 48-72 hours of life.

Newborn Screening samples are preferably collected by heel prick, but venous or arterial samples are acceptable.

Blood should be dropped directly onto the preprinted filter paper card. DO NOT USE lithium heparin or EDTA tubes as these anticoagulants interfere with the test results.

DO NOT USE Vaseline, paraffin etc on the heel as this can interfere with the test results.

• Requirements for capillary heel prick sample:
  ‣ Newborn screening card
  ‣ Gloves
  ‣ Alcohol/sterile water / no cleaning necessary if after a bath
  ‣ Sterile cotton wool swabs
  ‣ **Retractable sterile lancet** [OH&S requirement] max length 2.4mm
    (must comply with Health and Safety issues and must be fully retractable)
  ‣ Baby’s/mother’s file and special stamp for signatures in file
  ‣ Baby’s Personal Health Record (Blue Book)

• **Warm foot** (take sample after warm bath, use booties, foot warmer [kept in NUM’s office] or warm hands). **DO NOT USE** cloths moistened with hot water.

• **If possible place the baby’s leg** lower than rest of body

• **Clean area** with alcohol swab or water / no cleaning necessary if after a bath

• **Dry area** with sterile cotton wool

• **Puncture heel firmly once** with retractable sterile lancet (point <2.4mm) on inner or outer border of the heel
- Allow time for puncture to ooze and wipe away first drop of blood with sterile wool swab

- Gently massage above puncture site to encourage blood flow and drop free flowing blood onto one side of the filter paper only

- Completely fill each circle. Blood must soak through card to other side

- Apply gentle pressure to the heel until bleeding stops. It is preferable not to use elastoplasts

- Hold newly collected samples horizontal for about 20 seconds, so the blood remains even

- Cards should be completely dried horizontally, preferably in a drying rack designed for the purpose or laid flat with blood spots overhanging a surface so air can circulate. Drying should be at room temperature away from artificial heat or sunlight (and takes approximately 4 hours). The drying racks are obtainable from the NSW Newborn Screening Laboratory.

- Do not layer successive drops of blood

- Do not squeeze heel – Squeezing causes interstitial fluid to ooze out with the blood, contaminating the sample

- Avoid the heel rubbing on the card

- Do not let card near milk formulae, antiseptic solutions, lotions, water, urine, etc

CONTAMINATION ADVERSELY AFFECTS THE SAMPLE AND MAY LEAD TO MISSED DIAGNOSIS
GUIDELINE (4)
DOCUMENTATION OF INFORMATION

- Use ball point pen only – DO NOT USE pencil, ink or felt pens
- All information to be legible and written in capitals
- All information to be completed, especially the baby’s feed (cow’s-milk-based/soy-based etc) and relevant clinical information is of the utmost importance eg meconium ileus (MI), mother on thyroxine, twin to twin transfusion, sibling PKU or CF, family history of a metabolic disorder etc

NB: It is most useful to have a hospital label with the mother’s/baby’s information on the BACK of the card. However, this label must not TOUCH the blood spot and the name must match the name written on the front

- The name of the paediatrician or doctor in charge of the baby must be written on the card. This must be the name of a doctor who is prepared to initiate follow-up of a baby with an abnormal result, and is the doctor newborn screening will contact. The name of the obstetrician is not useful in most hospitals.

- Document in the mother’s/baby’s file that:
  - Pamphlet has been given
  - Contents of pamphlet discussed and verbal consent given
  - Verbal consent
  - Newborn screening test has been completed

- Pre-inked stamp or equivalent to be used similar to example below.
  All parts need to be completed.

<table>
<thead>
<tr>
<th>Baby's name:</th>
<th>Signature (Health Professional)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Provision of the NBS pamphlet:</td>
<td>Date:</td>
</tr>
<tr>
<td>Discussion of NBS information:</td>
<td>Date:</td>
</tr>
<tr>
<td>Verbal consent:</td>
<td>Date:</td>
</tr>
<tr>
<td>Completion of NBST test:</td>
<td>Date:</td>
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GUIDELINE (5)
SENDING SAMPLES

- **Alternate the blood spots** when stacking for posting so that the blood spots do not touch each other.

- **Wrap samples together** in a sheet of white paper or put in a paper bag – Australia Post will only transport wrapped samples in an envelope. **Approx. 8 cards will fit into a standard envelope.**

- **Post daily** or send by courier regularly.

NEVER PUT WET SAMPLES IN A PLASTIC BAG/GLADWRAP because this encourages bacterial growth and INVALIDATES test results.

SEND SAMPLES DAILY

| Postal Address: | NSW Newborn Screening Programme  
| Locked Bag 2012  
| WENTWORTHVILLE NSW 2145 |
| Courier Address: | NSW Newborn Screening Programme  
| The Children's Hospital at Westmead  
| WESTMEAD NSW 2145 |
| Telephone Contacts: | Programme Clinical Nurse Consultant  
| (02) 9845 3255  
| Programme Secretary  
| (02) 9845 3659  
| Fax:  
| (02) 9845 3800 |

SAMPLES MUST NOT BE BATCHED BUT:
POSTED DAILY OR
SENT BY COURIER REGULARLY
GUIDELINE (6)  
REFUSALS

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 must develop a protocol for parental refusal of a newborn screening test for their baby.

- **Parents given pamphlet** “Tests to Protect your baby”
- **Parents properly informed** by the midwife about the test and its importance
- **IF TEST REFUSED**
  - Paediatrician re-enforces importance of newborn screening
  - Parents are given the telephone number of newborn screening and the option of speaking to a senior officer of the programme for any further information
- **IF TEST STILL REFUSED** Parents sign the hospital’s disclaimer form (suggested disclaimer form attached to this guideline – attachment 4). This form is put in baby’s or mother’s hospital file
- **Midwife completes information** on newborn screening card, writes “REFUSAL” on it and sends card to newborn screening. (This is important for legal protection of both the hospital and the laboratory)
NSW NEWBORN SCREENING PROGRAMME
DISCLAIMER FOR BABY’S NEWBORN SCREENING TEST

NSW Health offers free newborn screening tests to all babies born in New South Wales and the Australian Capital Territory to detect rare metabolic disorders. A blood sample is ideally taken at 48-72 hours of age, by heel-prick, and sent to the NSW Newborn Screening Laboratory. About 90 babies each year are found with one of these rare disorders, which are treatable either by diet or medications or both.

The pamphlet “Tests to protect your baby” should be given to you, the contents discussed and your verbal consent given. If for any reason you are reluctant to let your baby have the tests, it is a good idea to discuss the implications with your paediatrician or telephone a senior officer of the NSW Newborn Screening Programme (Tel: 9845-3255 or 9845-3659) before you make your final decision.

If your final decision is that you do not wish your baby to have the tests, please sign the following disclaimer:

We have had the information brochure "Tests to Protect Your Baby" explained. We understand the risks that may be involved to the baby if the newborn screening tests are not performed.

We understand that neither the hospital and its staff nor the NSW Newborn Screening Programme can be held responsible for any consequences suffered by the baby as a result of not being tested.

Signed:

Mother: ___________________________ Print Name: ___________________________
Father:___________________________ Print Name:_________________________
Witness:_________________________ Print Name:_________________________

Date:_____/_____/_____

Attachment 4
GUIDELINE (7)
HOSPITAL NOMINATED NEWBORN SCREENING LIAISON PERSON:

In accordance with the Health Department Circular “Newborn Screening Guidelines – PD2005_273), hospitals should ensure that a nominated person (eg community liaison midwife, nurse unit manager of maternity etc) is responsible for newborn screening. The nominated person should have a relief newborn screening person for holiday/sick and other types of leave. The name and position of the nominated and relief person should be notified in writing to the NSW Newborn Screening Programme.

Responsibilities of the nominated newborn screening liaison person are:

- Act as the contact for NSW Newborn Screening Programme
- Check that all babies have been offered the test
- Check that babies have had samples taken
- Check the quality of blood samples and data on cards before arranging for cards to be sent daily by post or by courier
- Contact the parents when a resample is necessary and given them the fact sheets, which are provided by the laboratory with the request to resample
- Inform the baby’s doctor of the need and indication for the resample
- Ensure resample is collected from babies whose initial sample was <48 hours

IT IS EXPECTED THE HOSPITAL WILL AUTOMATICALLY SEND ANOTHER SAMPLE

- Check the confirmation report against the hospital births as soon as possible

The following actions are to be taken as necessary:

- 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 missed having a sample collected, arrange collection. The reason the baby was missed should be documented in the file
- Send any corrections such as name, date of birth, date of sample etc to the laboratory for update of the sample card and database

The hospital of birth is responsible for ensuring all babies are offered the newborn screening test, including babies who are transferred to another hospital
GUIDELINE (8)
COMMUNITY MIDWIVES PROGRAMMES:

1. Please apply Guidelines 1, 2, 3, 4, 5 & 6 with particular attention to the following:

1.1 After newborn screening sample has been taken:

either

• lay card flat with the actual blood spots overhanging: ie not touching the surface of shelf etc or
• place horizontal in drying rack in small esky. Punch holes in esky to allow for air circulation

DO NOT PUT FROZEN ICE BLOCK IN ESKY
DO NOT PUT SAMPLE IN PLASTIC

The sample must not be left in a hot car. (The heat in the car will make the results unreliable).

1.2 The following decisions could be made:

• Preferably take card in esky into each visit
• If your next visit is for not more than 30 minutes, leave esky in boot of car as it is slightly cooler
• If the parents seem reliable ask them to keep the card, wrap it, and post the next day when dry

1.3 Post or courier the sample as soon as possible
OTHER NEWBORN SCREENING ISSUES

1. Timing of sample collection

- Recommended time is when the baby is 48 – 72 hours old. Certain metabolic disorders may be missed if the baby is less than 48 hours old.
- If the baby is discharged before 48 hours old, the test must be carried out before the baby leaves the hospital unless the hospital of birth has a fail-safe community midwifery program. The repeat test may be collected in the community as soon after 72 hours as possible.
- The hospital of birth is responsible for the newborn screening tests.
- If the baby is moribund, the test must be carried out before the anticipated neonatal death. This allows testing which may significantly benefit the family.

2. Hospital transfers - babies who may have a delayed or missed screening test include:

- Sick neonates transferred from one hospital to another.
- Well neonates transferred from a major or base hospital to a district or country hospital.
- Neonates who are transferred to a community midwifery program.

Special care must be taken by hospitals and community programs to ensure that a sample is collected. The hospital protocol for transferring and/or receiving babies should be followed. A sample should be collected if possible before transfer to another hospital. The receiving hospital should check if another sample needs to be collected.

As previously stated it is the hospital of birth who is responsible for all babies being offered the newborn screening test and ensuring that it is taken.

3. Blood transfusions

- A newborn screening sample must be taken BEFORE any blood transfusion. If this does not occur, a sample should not be taken until 48 hours after a blood transfusion. Enzyme tests to confirm galactosaemia will be invalidated after any blood transfusion. DNA tests to confirm cystic fibrosis or other metabolic disorders will be invalidated for 6 months after an exchange blood transfusion.

- Where a blood transfusion has occurred, write in “relevant clinical information”:
  - date of blood transfusion
  - time of blood transfusion
4. Parenteral nutrition (TPN/IVI etc)
   - Note on card – TPN can alter test results

5. Arterial Lines
   - A blood sample may be taken from an arterial line. Approximately 1ml should be removed from the line and saved, the sample then taken and the saved blood replaced - (Each hospital should have their own protocol)

6. Established feeds
   - Milk feeding is not required before newborn screening sample is collected regardless of weight and maturity of baby, but accumulation of metabolites and interpretation of results is easier after at least one feed
   - The feeding status of the baby at time of sample must be clearly stated on the card eg: “breast” / “cows-milk-based formula” / “soy-based formula” / “TPN” / “other”
   - If there are clinical indications of galactosaemia and the baby is on TPN or soy-based milk please notify the laboratory, so that the need for additional tests can be evaluated

7. Twin-to-twin transfusion
   It is possible to miss a metabolic disorder in a twin who had twin-to-twin transfusion. It is essential that this information be written on the card. A request for a repeat sample when twins are aged 4 weeks will be sent.

8. Stillbirths / Neonatal Deaths
   - For completeness of records, data collection and legal purposes please return cards from:
   - Stillbirths – complete details and mark card stillbirth

STILLBIRTH DEFINITION IS: The complete expulsion or extraction from its mother of a product of conception of at least 20 weeks gestation or 400 grams birth weight who did not, at any time after delivery, breathe or show any evidence of life such as a heart beat.

- Neonatal Deaths - If the baby dies unexpectantly, complete details and mark card NND

NEONATAL DEATH DEFINITION IS: The death of a live born infant within 28 days of birth. Collection of a newborn screening sample prior to death is required IF a neonatal death is anticipated.

DO NOT RETURN A CARD TO NOTIFY THAT A BABY HAS BEEN TRANSFERRED TO ANOTHER HOSPITAL
9. Sample Collection Problems

- **Alcohol/water dilution** - if alcohol/water is not wiped away with a sterile cotton wool swab, it will dilute the sample and adversely affect a test result
- **Incorrect puncturing** - Puncturing the heel at sites other than those recommended may predispose to bony injury and possible infection. **ONLY PUNCTURE AROUND OUTER OR INNER BORDER OF HEEL** (see diagram page 11)
- **Layering** - Layering successive drops of blood on the same circle spot could affect the test result
- **Touching** – Avoid the baby’s heel rubbing the card
- **Squeezing causes haemolysis** - Squeezing the baby’s heel too hard may cause interstitial fluid to ooze out with the blood and contaminate the sample. *If blood flow diminishes and circles are not filled, repeat the puncturing technique*

10. Test Results

Results are usually available within two working days after receipt of sample.

- **Urgent Follow-Up**
  The paediatrician / doctor / independent midwife shown on the newborn screening card is notified by telephone of test results which are clearly abnormal and could indicate any one of the disorders. It is the responsibility of this person to ensure that the baby is promptly referred for further investigation and treatment.

- **Retesting**
  Retesting is carried out in the following circumstance:
  
  - Moderately abnormal results, which are likely to be transient but require confirmation – a written request for resample is sent by the laboratory
  - Insufficient / contaminated test samples – a written request for resample is sent
  - Twin to twin transfusions – a written request for a resample at 4 weeks is sent

  *When blood is collected before 48 hours, it is expected the hospital will automatically send another sample collected as soon as possible after 48 hours.*

11. Babies From Overseas

- **Babies arriving in Australia** up to age of 1 year, who have not had a newborn screening test in their country of birth, may be offered the newborn screening test, and results will be interpreted according to the age of the baby