**MORE ABOUT SCREENING**

**THE SAFETY OF THE SCREENING PROCESS**
Screening is quick and safe. Because the heel prick causes a small break in the skin, there is a small risk of infection. The midwife or nurse collecting the sample will use gloves and clean the heel before the test. You may wish to breastfeed or hold your baby during the test as the heel prick may cause a little discomfort to your baby.

**The Newborn Bloodspot Screening Process**

<table>
<thead>
<tr>
<th>Process</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Baby born</td>
<td>48-72 hours</td>
</tr>
<tr>
<td>Midwife/nurse does screening</td>
<td>Midwife/nurse does screening</td>
</tr>
<tr>
<td>Card sent to laboratory for testing</td>
<td>Result negative</td>
</tr>
<tr>
<td>Result positive</td>
<td>Storage min 2 years - 18 years</td>
</tr>
<tr>
<td>Result negative</td>
<td>Result negative</td>
</tr>
<tr>
<td>Result positive</td>
<td>Referred to specialist</td>
</tr>
<tr>
<td>Referred to specialist</td>
<td>Result negative</td>
</tr>
<tr>
<td>Storage 2 years - 18 years</td>
<td>Result positive</td>
</tr>
</tbody>
</table>

**WHAT IS A NEGATIVE RESULT?**
A negative screening result means your baby has not been identified as having one of the conditions being screened. On very rare occasions, a false negative may occur where the baby has a normal screen but develops symptoms for one of the conditions later. This occurs in approximately one in 100,000 cases.

**WHAT IS A POSITIVE RESULT?**
A positive screening result does not necessarily mean your baby has a particular condition. Newborn screening identifies babies at increased ‘risk’ of a condition. Further testing is needed to confirm the result. Babies with a positive screening result have more samples collected. If the later screens are also positive, your baby will be referred to a specialist.

**DNA TESTING**
Newborn bloodspot screening involves biochemical testing not DNA testing. However, approximately one per cent of babies from the biochemical testing will show a risk for cystic fibrosis or a fatty acid oxidation disorder which will then be DNA tested. No DNA tests are done on any other samples.

**HAVING A LATER SCREENING**
If you choose not to have your baby screened and you change your mind later, speak to your family doctor. Your doctor will arrange for your baby to have the right tests.

**MORE INFORMATION**

**NEWBORN BLOODSPOT SCREENING PROGRAM**
Address: Locked bag 4001, Wentworthville, NSW 2145
Tel: 61-2-98453659
Email: NSWH-newbornscreening@health.nsw.gov.au

**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 or midwife may need to follow up your baby’s test results with further investigations.

**ORDER ADDITIONAL–NEWBORN BLOODSPOT SCREENING BROUCHURES** from NSW forms and brochure ordering system.
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Locked Mail Bag 861, North Sydney 2059
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**TESTS TO PROTECT YOUR BABY**
**EARLY DIAGNOSIS IS IMPORTANT TO YOUR BABY’S FUTURE HEALTH AND WELLBEING**
Newborn Bloodspot Screening is a free blood test that is offered to every newborn baby in NSW and the ACT. This blood test is to check if your baby may have been born with a medical condition.

If your baby is not screened at birth and has one of the conditions being screened, the baby’s development may already be affected by the time symptoms appear. While a baby will be offered treatment when symptoms appear, their growth and development could already be affected. Some conditions that are screened for may be life threatening if untreated. Early screening and detection means treatment can start early.

When Screening Occurs
When your baby is between two and three days old (that is between 48 and 72 hours old), your baby will have the test. A midwife or nurse will prick your baby’s heel with a lancet (a special needle) and will collect a few drops of blood onto a screening card. Then, the card is sent to a central laboratory for processing.

CONSENT FOR SCREENING
Before a sample is collected, you must give NSW Health your signed consent that you agree to the screening. There is a consent section on the screening card (see below) which you will be asked to sign if you want to go ahead with the screening.

NSW NEWBORN SCREENING PROGRAMME
Consent for Collection and Testing of Sample
I have read and understood the information in the NSW Newborn Screening pamphlet. I consent to my baby having blood collected and tested

Tick ‘Yes’ or ‘No’:
1. Consent to have your baby screened.
2. Consent to store the screening card for more than 2 years.
3. Consent for the card to be used for personal identification.

Storage of screening card for greater than 2 years
I consent to the storage of the screening card for longer than 2 years

Consent on the screening card
You can choose to say YES to the first and NO to the other two items if you wish.

Screening for Particular Medical Conditions
The Newborn Bloodspot Screening Program screens approximately 100,000 babies per year for 25 medical conditions. From time to time, further disorders will be added to or removed from the screening program. The following are the most commonly diagnosed conditions:

Primary congenital hypothyroidism
• An absence or abnormal formation or function of the thyroid gland affects growth and causes intellectual disability if untreated.
• Around 40 babies per year are diagnosed.
• Treatment is to medicate with thyroid hormone.

Cystic Fibrosis
• A dysfunctional gene results in thick mucus in different organs throughout the body, which leads to severe chest infections and a failure to thrive if untreated.
• About 30 babies per year are diagnosed.
• Individuals with cystic fibrosis have a great improvement in their health if they start treatment early.

Phenylketonuria (PKU)
• The body is unable to breakdown the essential amino acid phenylalanine and can lead to severe intellectual disability if untreated.
• About 10 babies per year are diagnosed.
• The treatment for PKU is a diet low in phenylalanine started in the first two to three weeks of life.

Medium Chain Acyl CoA Dehydrogenase (MCAD) Deficiency
• Inability of the body to completely break down fat. If untreated, it may be life-threatening during common childhood illnesses.
• About 6-8 babies a year are diagnosed.
• Treatment involves taking extra precautions during illnesses to ensure the child gets adequate energy intake.

Congenital Adrenal Hyperplasia
• Altered ability of the adrenal gland to produce hormones that may affect the baby’s metabolism, response to infection, ability to regulate salt levels and sex characteristics.
• About 6-7 babies a year are diagnosed.
• The treatment is through medication such as hormone replacement and salt replacement.

Other rare disorders
• There are other rare disorders that may affect babies: approximately 20 per year are diagnosed by the NSW Newborn Screening Program.