Phenylketonuria
(PKU)

Congenital Hypothyroidism

And

Sickle cell Disorder

ABOUT YOUR BABY'S BLOOD TEST

What is it?

This is a simple blood test, which is performed by your Midwife between seven and ten days after birth. It is also known as the Guthrie Test
It is a screening test and is available for every baby to identify two conditions: Phenylketonuria and Congenital Hypothyriodism. These conditions cannot be detected by normal examination of the newborn baby and if not detected early can harm the baby's brain causing retardation.

Why is it done?

To identify:

1. Phenylketonuria (PKU)
   PKU affects 1 in 10,000 babies. These babies cannot use utilise part of a protein in their food resulting in a build up in the blood which can lead to brain damage unless corrected.

2. Congenital Hypothyroidism
   Affect 1 in 4000 babies and is caused by underactivity or absence of the thyroid gland in the baby's neck. This gland doesn't produce enough of the hormone thryroxine, which is essential for normal growth and brain development.

How is it done?

The midwife will obtain a sample of blood from the baby's heel using a small needle when the baby is between seven and ten days old. The sample is collected on a special absorbent card and may be performed in hospital or at home.

What happens to the blood sample?

It is sent to the laboratory responsible for carrying out the tests in your area. Occasionally a repeat blood sample is requested for technical reasons or because the baby was small at birth. A repeat test does not necessarily

indicate that something is wrong. The test results are usually available within 2 weeks

How will I know the results?

If the results are normal you are not informed routinely. However, your Health Visitor or GP will be informed.

What if the results are positive?

Your Midwife or GP will contact you and arrangements will be made for you to attend the local hospital where further investigations will be carried out and decide whether or not your baby needs treatment.

How is it treated?

In the case of PKU, this will involve the introduction of a special feed to replace infant formula. Breastfeeding can be continued if desired and the baby is eventually weaned onto a lifelong diet.

In the case of Hypothyroidism, tablets (or powders, thyroxine is not available in powder or liquid form only tablet***)*** which replace the missing hormone will be prescribed for your baby. No special diet is necessary.

Sickle Cell Disorder

Sickle cell disorder is an inherited disorder of the red blood cell which may be life threatening. It affects 1 in 2380 babies.
The disorder can only occur if one parent carries sickle cell trait, and the other carries another significant haemoglobin trait (e.g. sickle cell trait, 'C' trait, beta Thalassaemia trait). If the condition is detected early, treatment can be given to reduce symptoms.

The screening test may also detect other inherited disorders of red blood cell such as Thalassaemia major. Again, early treatment can be given to reduce symptoms.

Babies who are born with a trait (healthy carriers) may also be identified by this screening test. This can provide useful information for them in the future.

What if I want to know more?

If you wish to know more about these tests, you should ask your Midwife and she will supply you with more information.

Although it is worrying to think about these disorders affecting your baby, it is highly unlikely that the test will be positive, but if so early detection and treatment will prevent the harmful effects and allow your baby to develop normally.

For further information regarding Phenylketonuria, please contact your midwife or;

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A useful web site address for both conditions is

www.cafamily.org.uk

This leaflet can be made available in other formats on request

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