Appendix 2

Information for parents when the bloodspot test has not been undertaken

The aim of the newborn bloodspot screening programme is to identify specific conditions, as soon after birth as possible and before the onset of clinical symptoms

Newborn bloodspot screening can identify the following conditions:

- Phenylketonuria (PKU)
- Congenital Hypothyroidism (CHT)
- Cystic Fibrosis (CF)
- Sickle Cell Disorders (SCD)
- Medium Chain Acyl CoA Dehydrogenase deficiency (MCADD)
- Glutaric Aciduria type 1 (GA1)
- Homocystinuria (HCU)
- Isovaleric Acidaemia (IVA)
- Maple Syrup Urine Disease (MSUD)

Screening allows these conditions to be detected early – usually before signs and symptoms become apparent. Early treatment can help prevent brain damage, serious illness, or in some cases death, in the case of PKU, CHT, GA1, HCU, IVA, MSUD, and MCADD. Early treatment can also limit the effects of CF and SCD. Delays in diagnosis may be detrimental to your child's health.

Because your baby has not been screened you should look out for any of the symptoms listed below. If you are concerned, please contact your GP for advice. Let them know that your baby has not been screened for one or more of the above conditions. By the time you notice anything is wrong it may be too late to prevent some lifelong damage but treatment, if started as early as possible, may still be of some benefit

If you wish your baby to be screened for any or all of the conditions please contact your GP or Health Visitor immediately. Testing is best carried out as early as possible. Screening becomes less reliable for CF after the baby is six weeks old although other tests can be used to diagnose CF at a later stage.

Symptoms and signs of Congenital Hypothyroidism (CHT)

- Constipation
- Dry skin
- Hoarse cry
- Large tongue
- Swelling around the eyes
- Feeding problems
- Sleeping for long periods
- Prolonged jaundice
- Baby fails to thrive and develop

Signs and Symptoms of Cystic Fibrosis (CF)

- Recurrent Infections of the lungs and sinuses
- Chesty cough that doesn't clear up
- Dirty nappies smell much worse than normal and are pale and greasy
- Poor growth and poor weight gain
- Salty taste to the skin

Signs and Symptoms of Sickle Cell Disorder (SCD)

- Increased susceptibility to infection
- Painful swelling of the fingers and hands or toes and feet. Pain can also occur in the arms, legs, back and abdomen (from about 6 months)
- Pale skin and lips
- Yellow pigment in the eyes and skin (jaundice)

Signs and Symptoms of Phenylketonuria (PKU)

- Baby fails to progress in learning to sit, use the hands, crawl and develop speech
- Unusual behaviour, such as screaming episodes, repetitive rocking, head banging or arm biting
- Vomiting leading to weight loss
- Irritability / crying a lot
- Skin problems, such as sensitivity to light, dry skin or itchy rashes
- Baby grows and develops more slowly than normal
- An unusual smell to the skin, hair and urine
- Fits / seizures

Phenylketonuria after childhood

There is a rare mild form of PKU, which causes little obvious effect, apart from mild learning difficulties. Boys and girls can reach adulthood and be unaware that they have this form of PKU. However if your child is a girl it is particularly important that she is aware that she has not been tested for PKU. If your daughter does have PKU, this may harm the development of any children she may have. She needs to know that she has not been tested for PKU to allow her to make choices about her future pregnancies. She may choose to be tested for PKU. Children born to boys with PKU are not affected in this way.

Signs and symptoms of Medium Chain Acyl CoA Dehydrogenase deficiency (MCADD)

Signs and symptoms can develop quickly in infants who are not feeding well or who are unwell with an infection or illness such as diarrhoea and vomiting

- Drowsiness
- Seizures
- Coma

Signs and symptoms of Glutaric Aciduria type 1 (GA1)

In children with GA1, a minor illness, such as a chest infection or a tummy upset, can lead to serious problems. Early signs may be:

- Vomiting
- Irritability
- Excessive sleepiness
- Floppiness
- Breathing difficulties

Signs and Symptoms of Homocystinuria (HCU)

- Learning difficulties
- Eye problems
- Bones which are abnormally long and thin (osteoporosis)
- Blood clots / strokes

Signs and Symptoms of Isovaleric Acidaemia (IVA)

Children with IVA can become severely unwell. Early signs may be:

- Vomiting
- Excessive sleepiness
- Floppiness
- Rapid breathing

Signs and Symptoms of Maple Syrup Urine Disease (MSUD)

Many babies with MSUD become unwell when they are a few days old, with:

- poor feeding
- vomiting
- excessive sleepiness
- Coma or permanent brain damage

In older children a minor illness, such as a chest infection or a tummy upset, can lead to serious problems. The early signs in older children include confusion and poor balance as well as loss of appetite, vomiting and excessive sleepiness. As with babies, this can lead to a coma unless treated correctly