

## NEWBORN BLOOD SPOT SCREENING

Newborn blood spot screening is undertaken shortly after birth to identify babies who may have been born with rare and serious conditions. In BFG, babies are already offered an extensive screening programme, combining most of England's and Germany's routine newborn screening programmes.

From 3 August 2009, screening for Cystic Fibrosis (a common inherited condition that affect the lungs and the pancreas) and Sickle Cell Disorders (an inherited cell disorder that affect the red blood cells, mainly in people of African, Caribbean, Mediterranean, Middle Eastern or Asian origin) will also be universally offered to babies born in BFG.

Newborn blood spot screening in Germany is undertaken after the baby is 36 hrs old. This may be in the hospital or by the community midwife at home. The midwife will collect some drops of blood from the baby's heel, using a special device to collect some drops of blood onto a card. Parents may know this as the heel prick test.

In the majority of cases, all screening tests return normal results. But if the baby is affected by a condition, the benefits of early identification can be enormous in improving their health and preventing serious illness. It also means parents will be given the appropriate care and guidance to help their child and help plan for the future.



### **Below is the list of conditions screened for**

#### **Adrenogenital Syndrome**

Hormone disorder because of a deficiency of the adrenal cortex: Masculinization in females, possible fatal progression due to salt depletion. Treatment with hormones (frequency approx 1/10,000 newborns)

#### **Maple Syrup Disease**

Deficiency in the depletion of amino acids: Mental handicap, coma, possible fatal progression. Treatment by means of a special diet (frequency approx 1/200,000 newborns).

#### **Biotinidase Deficiency**

Deficiency in the metabolism of the biotin vitamin: Skin changes, metabolic crisis, mental handicap, possible fatal progression. Treatment by means of administering biotin. (Frequency approx 1/80,000 newborns).

#### **Carnitine Metabolic Deficiency**

Metabolic deficiency of fatty acids: Metabolic crisis, coma, possible fatal progression. Treatment by means of a special diet. (Frequency approx 1/100,000 newborns).

#### **Galactosemia**

Metabolic deficiency of lactose: Loss of sight, physical and mental handicap, liver failure, possible fatal progression. Treatment by means of a special diet. (Frequency approx 1/40,000 newborns).

#### **Glutaric Aciduria Type I**

Deficiency in the depletion of amino acids: Permanent motoric disturbance, sudden metabolic crisis. Treatment by means of a special diet and administering amino acids. (Frequency approx 1/80,000 newborns).

**Hypothyreosis/Hypothyroidism**

Hereditary sub-function of the thyroid: Severe disturbance of physical and mental development. Treatment with hormones. (Frequency approx. 1/4,000 newborns).

**Isovalericacidemia**

Deficiency in depletion of amino acids: Mental handicap, coma. Treatment by means of a special diet and administering amino acids. (Frequency approx 1/50,000 newborns).

**LCHAD-, VLCAD Deficiency**

Metabolic disorder of long-chain fatty acids: Metabolic crisis, coma, muscle and cardiac insufficiency, possible fatal progression. Treatment by means of a special diet, avoiding periods of starvation. (Frequency approx 1/80,000 newborns).

**MCAD Deficiency**

Deficiency of energy recovery from fatty acids. Metabolic crisis, coma, possible fatal progression. Treatment by means of administering carnitine, avoiding periods of starvation. (Frequency approx 1/10,000 newborns).

**Phenylketonuria**

Metabolic deficiency of the amino acid phenylalanine: Fits, spasticity, mental handicap. Treatment by means of a special diet. (Frequency approx 1/10,000 newborns).

**Sickle Cell Disorders (includes Thalassaemia)**

Red blood cell disorder: Blocked capillaries, stops oxygen to tissues, pain, anaemia, infection susceptibility. Treatment with antibiotics and vaccines to reduce the risk from serious infections. (Frequency in UK overall 1 in 2,400. Most common in people of Afro Caribbean or sub Saharan origin. Also found in Arab, Mediterranean and Indian origins as well as others).

**Cystic Fibrosis (CF)**

Condition affects certain organs in body particularly pancreas and the lungs. Thick secretions in these organs cause digestive problems and chest infections. (Frequency in UK - equivalent to 1 in 2,500 babies. *Carriers*: 1:24 Caucasians, 1:55 UK residents of Asian origin, 1:70 UK residents of Indian sub-continent origin). Screening results are not 100% conclusive. Treatment includes antibiotics and physiotherapy.

**Note:**

*Not all above-named illnesses can be completely prevented even if treatment commences early. A prompt treatment in most cases enables the affected child to develop normally.*

Although the screening of babies is strongly recommended, it is not compulsory and parents should ensure they understand the purpose and possible results of all the tests before they choose to take them. During routine antenatal care, parents are advised to discuss newborn screening tests with their community midwife, health visitors or GP.