Neonatal Screening Test (Heel Prick Test)

Your baby will be offered a Screening blood test to detect and identify some rare but serious diseases. Early detection and treatment can improve health and prevent severe disabilities.

WHAT CONDITIONS ARE BEING TESTED FOR?

- *Cystic Fibrosis (CF)* is a serious inherited condition that affects approx. 1:2500* babies. Some of the problems associated with CF are poor digestion and chest infections. Early diagnosis means that measures can be taken to prevent infections and improve quality of life.

- *Congenital Hypothyroidism* affects approx. 1:3000* babies. With this condition babies do not produce enough of the hormone, thyroxine, which is vital for normal mental and physical development. It is treated by giving thyroxine tablets which will help normal development.

- *MCAD-Deficiency* affects approx. 1:8000* babies. It is an inherited disorder where the body is unable to digest fatty acids beyond a certain point, if early detected it can be managed by adjusting the diet, avoid fasting and illness.

- *Phenylketonuria (PKU)* affects approx. 1:10.000 babies. These babies cannot digest a substance called phenylalanine, which is present in protein-rich foods such as milk. A build up of this substance can result in severe brain damage. If PKU is detected early, the baby is given a special diet, which excludes phenylalanine, and allows the baby to develop normally.

- Glutaric aciduria type 1(GA1) affects about 1:100,000 babies worldwide and they are unable to break down amino acids known as lysine, hydroxylysine and tryptophan. Without treatment, the child can go into a coma and are at risk of brain damage. The treatment consists of a special low protein diet and medicine which help to prevent the build-up of harmful substances in the blood whilst ensuring that the child receives enough protein to grow and develop.

- Maple syrup urine disease (MSUD) affects about 1:185,000 children worlwide. They will have problems breaking down particular amino acids known as leucine, isoleucine and valine. Without treatment, this leads to a coma and they will also be at risk of brain damage but with the correct treatment the outcome can be greatly improved.

- Homocystinuria (HCU) affects about 1:250,000 babies and they are unable to break down amino acid known as methionine which causes a chemical called homocysteine. Without treatment most children have vision problems, may also develop osteoporosis and are at risk of blood clots or strokes. In some children the level of homocysteine can be controlled by giving Vitamin B6, if it does not work the treatment is a special low protein diet and extra supplements and medicine.

- Isovaleric acidaemia (IVA) affects about 1:250,000 babies and they are unable to break down the amino acid leucine. Without treatment severe life threatening symptoms can occur, such as, seizures or comas. With early diagnosis and early treatment babies with IVA will be able to live healthy lives. The treatment consists of a special diet and medicines.

*rate in the UK

HOW, WHEN AND WHERE IS IT DONE? BY WHOM?

The test is taken when baby is between 5-8 days old, with day 5 being the optimal day and it will usually be the Community Midwife who performs the test at a clinic appointment. They will take several drops of blood from a small "prick" on baby's heel to fill up 4 circles on a special absorbent card. This card is then sent to a laboratory in the UK. You can help settle your baby by cuddling or feeding them whilst it is being performed.

WHAT HAPPENS TO THE RESULTS?

The results are usually returned to the Maternity Unit 3-4 weeks after being sent.

Negative results – You will not be contacted if the results are normal. **No News is Good News!**

Positive results – You will be contacted by the Maternity Unit to make arrangements for further testing or necessary appointments

WHY ARE TESTS SOMETIMES REPEATED?

- Not enough blood was taken to allow proper testing.
- The blood card was damaged or did not reach the laboratory.
- Test results were inconclusive.

WHERE CAN FURTHER INFORMATION BE OBTAINED?

National Society for Phenylketonuria www.nspku.org

British Thyroid Foundation www.btf-thyroid.org

Cystic Fibrosis Trust www.cftrust.org.uk

CLIMB (National Information Centre for metabolic diseases) www.climb.org.uk

For any further information please contact :

The Maternity Unit at St. Bernard's Hospital on:
+350 20072266 ext: 2125

gha
Gibraltar Health Authority