HCU: further information

This information is for parents if their baby is suspected of having HCU or has been diagnosed with HCU following their newborn blood spot screening test ('heel prick test'). It will help you and your health professionals to talk through the next stages of your baby's care.

About HCU

Homocystinuria (pronounced ho-mo-sistin-ur-ee-a), or HCU, is a rare but treatable inherited metabolic disorder that prevents the normal breakdown of protein. Babies with HCU inherit 2 faulty copies of the gene for HCU, one from each parent.

When we eat, our body breaks down protein in food into smaller parts called amino acids. Enzymes, which are chemicals found naturally in our body, then break down the amino acids further so they can be used.

In HCU, an enzyme called cystathionine beta-synthase is missing, which leads to problems breaking down the amino acid methionine. This causes methionine and a harmful substance called homocysteine to build up in the body and blood.

Untreated HCU can cause long-term health problems, including learning and behavioural difficulties, problems with vision, bone abnormalities, and an increased risk of blood clots and strokes. However, with newborn screening and life-long treatment, these can be prevented.

Screening and diagnosis of HCU

Newborn blood spot ('heel prick test')

When your baby was about 5 days old, your midwife took some blood from your baby's heel for their newborn blood spot screening test (the 'heel prick test'). The newborn blood spot screening test measures the amount of some amino acids in the blood. A high level of the amino acids methionine and total homocysteine suggests your baby may have HCU. This is called a screen positive result.

Diagnostic tests

If your baby has a screen positive result, you will be seen by a metabolic doctor, dietitian and nurse specialist (the 'metabolic team'). The team will provide advice and support. Blood tests will be carried out to confirm or exclude the diagnosis of HCU.

You will need to wait a few days for the test results. During this time, you can continue to breastfeed or give normal infant formula.

Treatment

If your baby does have HCU, the metabolic team will:

- explain the condition in detail and answer any questions you might have
- start a trial of treatment using vitamin B6 (pyridoxine)
- provide you with information about medical and dietetic treatment
- provide you with written information and contact details for the metabolic team
- arrange regular follow-up appointments

The metabolic team will see you regularly throughout your child's life and your child will remain under their care. Your baby will need regular blood tests to monitor the levels of homocysteine in their blood.

Vitamin B6 (pyridoxine) treatment

In some babies, the blood homocysteine levels can be lowered by giving vitamin B6 (pyridoxine). Your baby will start a trial of vitamin B6 (pyridoxine) to see if this reduces their homocysteine to a normal level.

The restricted protein feeding plan

If the vitamin B6 (pyridoxine) does not work, a low-methionine feeding plan will be started. The aim of the feeding plan is to reduce the build-up of homocysteine, which is formed from the amino acid methionine.

Your baby will be given 2 different feeds to provide all the nutrition they need for growth and development. These are:

- a limited daily volume of breastmilk feeds or infant formula
- a low-methionine infant formula

The level of methionine and total homocysteine in your baby's blood will be monitored on a weekly or fortnightly basis during the first year. Your metabolic dietitian will teach you how much of each feed to give based on the blood test results and your baby's weight. Following the dietetic instructions is very important for your baby's health.

Your GP will be asked to prescribe the low-methionine infant formula. You will continue to have regular appointments with a metabolic dietitian who will advise on dietary management throughout the various stages of childhood.

Other treatment

In later childhood, a medication called betaine is sometimes given to help reduce blood homocysteine levels if they are not in the treatment range.

Long-term outlook

With life-long treatment and well controlled homocysteine levels, the outcome is usually very good and most children will avoid any long-term health problems.

Your other children

At-risk siblings

Children from the same parents have a 1 in 4 chance of having the condition. Your other children might be at risk of HCU even if they have never shown any symptoms. It is therefore very important to get them tested if they have not been previously screened for HCU. Your metabolic team will be able to arrange this testing.

Future children

A new baby from the same parents will also have a 1 in 4 chance of having HCU. When you find out that you are pregnant, you should tell your midwife and GP there is a family history of HCU. You should also inform your metabolic team early in the pregnancy.

The metabolic team will write a birth plan for you. This will include advice on an early screening test for your new baby which should be taken between 24 and 48 hours after birth. This blood test will be in addition to the routine newborn blood spot screening test. The birth plan will be given to you and shared with your obstetrician and local midwifery team.

You should notify your metabolic team once you have given birth so they can ensure the correct blood tests are taken and sent to the screening laboratory without delay.

Confidentiality

The NHS screening programmes use personal information from your NHS records to invite you for screening at the right time. Public Health England also uses your information to ensure you receive high quality care and to improve the screening programmes. Find out more about how your information is used and protected, and your options at www.gov.uk/phe/screening-data.

More information and support

The metabolic team will be happy to discuss any queries you might have.

Further information can be found at Metabolic Support UK at www.metabolicsupportuk.org.The Metabolic Support UK team can be contacted at:

- 0845 241 2173 or 0800 652 3181
- contact@metabolicsupportuk.org

The British Inherited Metabolic Diseases Group (BIMDG) website has a TEMPLE booklet about HCU at www.bimdg.org.uk/site/temple.asp.

NHS.UK has information about HCU at https://www.nhs.uk/conditions/homocystinuria/ and newborn blood spot screening at https://www.nhs.uk/conditions/baby/newborn-screening/blood-spot-test/.

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