HCU: an overview

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-sis-tin-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.

Untreated HCU can lead to long-term health problems, including visual problems and learning difficulties, but with newborn screening and early treatment this can be prevented.

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 more detail and answer any questions you might have. They will start your baby on medical treatment and a special diet, and they will arrange regular follow-up appointments. Following the diet and treatment instructions is very important for your baby's health.

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.

For more information, see Homocystinuria (HCU): further information at www.gov.uk/government/publications/hcu-confirmed-diagnosis-description-in-brief.

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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