Phenylketonuria (PKU): summary

Public Health England (PHE) created this information on behalf of the NHS. In this information, the word ‘we’ refers to the NHS service that provides screening.

This information is for parents if their baby is suspected of having PKU or has been diagnosed with PKU 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 PKU

Phenylketonuria (pronounced as fee-nile-key-tone-you-ree-ah), or PKU, is a rare but treatable inherited metabolic disorder that prevents the normal breakdown of protein. Babies with PKU inherit 2 faulty copies of the gene for PKU, one from each parent.

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

Screening and diagnosis of PKU

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 acid phenylalanine suggests your baby may have PKU. This is called a screen positive result, which you can see more about at https://www.gov.uk/government/publications/nhs-population-screening-glossary-of-terms/glossary-of-terms#screen-positive.

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 if your baby has PKU.

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

Treatment

If your baby does have PKU, the metabolic team will explain the condition in more detail and answer any questions you might have. They will start your baby on a special diet (to restrict phenylalanine intake), and they will arrange regular follow-up appointments.

Following the diet 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.

Find out how to opt out of screening at www.gov.uk/phe/screening-opt-out.

More information and support

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

For more information on phenylketonuria, go to www.gov.uk/government/publications/pku-confirmed-diagnosis-description-in-brief.

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

Phone: 0845 241 2173 or 0800 652 3181

Email: contact@metabolicsupportuk.org

The National Society for Phenylketonuria (NSPKU) also provides information at http://www.nspku.org/ and support at:

Phone: 030 3040 1090

Email: info@nspku.org

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

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

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