Phenylketonuria (PKU): detailed information

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-keytone-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.

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 PKU, an enzyme called phenylalanine hydroxylase is missing, which leads to problems breaking down the amino acid phenylalanine. This causes harmful levels of phenylalanine to build up in the brain and blood.

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 detail and answer any questions you might have
- start your baby on a restricted protein (low in phenylalanine) feeding plan
- teach you how to do home blood tests (which you will need to post to the metabolic laboratory for them to monitor the levels of phenylalanine)
- 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.

The restricted protein feeding plan

The aim of the feeding plan is to reduce the build-up of phenylalanine.

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 phenylalanine-free infant formula

The level of phenylalanine in your baby's blood will be monitored on a weekly 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 phenylalanine-free 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.

Long-term outlook

With good dietary treatment and well controlled blood phenylalanine 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 PKU even if they have never shown any symptoms. Your metabolic team will discuss if testing is necessary.

Future children

A new baby from the same parents will also have a 1 in 4 chance of having PKU. When you find out that you are pregnant, you should tell your midwife and GP there is a family history of PKU. 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 48 and 72 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.

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/newborn-screening/blood-spot-test/.

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