Sgrinio Smotyn Gwaed Newydd-anedig Cymru
Newborn Bloodspot Screening Wales

GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

Newborn bloodspot screening programme

Your baby's screening result

Phenylketonuria (PKU) is suspected

Your baby's screening result

The result of your baby's 'heel prick' screening blood test suggests they might have phenylketonuria (PKU) or a related condition. Your baby now needs further tests to check whether he or she has PKU or not.

This leaflet gives you some information about PKU and explains what happens next.

What is PKU?

Phenylketonuria (PKU), pronounced as fee-nile-key-tone-you-ree-ah, is a rare but treatable inherited disorder that prevents the normal breakdown of protein.

Babies with PKU inherit two 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. Special chemicals found naturally in our body, called enzymes, then make changes to the amino acids so our body can use them.
Babies with PKU do not have one of the enzymes that help break down one of the amino acids. This causes a harmful substance called phenylalanine to build up in their blood and urine.

Babies with PKU benefit significantly from early treatment and can live healthy and active lives. Without early diagnosis and treatment, babies with PKU can develop long-term health problems, including learning difficulties.

Treatment

PKU can be treated with a special low-phenylalanine diet and dietary supplements.

The special diet prevents the build-up of phenylalanine in the blood. These levels are monitored and the child's diet adjusted as needed. Dietary treatment for babies with PKU should start as early as possible and continue throughout life.

Related conditions

Some babies have phenylalanine levels in the blood that are only slightly higher than normal. These babies might not need any dietary treatment. They might only need to have their phenylalanine levels monitored by blood tests.

This condition, sometimes known as hyperphenylalaninaemia, is less common than PKU. The specialist doctor will be able to give you more information when you see them.

What happens next?

You will be given an appointment to see a specialist metabolic team who will:

- discuss the screening test result with you
- arrange blood and urine tests for your baby, and
- explain how these tests can confirm if your baby has PKU.

If PKU is confirmed, your specialist metabolic team will:

- explain how to give your baby the special low-phenylalanine diet (if your baby is breastfed, you can continue breastfeeding)
- give you any special dietary supplements and medications your baby will need
- give you written information about PKU for you to share with your family, GP and local hospital
- answer any questions you might have, and
- arrange a follow-up appointment to discuss the test results.

Until you see your specialist metabolic team, you can breastfeed or bottle-feed your baby in the usual way.

If you are concerned about poor feeding or that your baby is unwell, contact your specialist metabolic team.

More information and support

- National Society for Phenylketonuria (NSPKU)
  Phone: 030 3040 1090 (helpline)
  Email: info@nspku.org
  Website: www.nspku.org

- Newborn Bloodspot Screening Wales:
  phw.nhs.wales/newborn-bloodspot-screening

Contact details for your specialist metabolic team

Specialist centre	
Consultant	
Metabolic dietitian	
Clinical specialist nurse	
Ward (if this applies)	

Notes

You can write down any notes here from discussions with your health professionals.

Using your information

For us to contact you as part of the programme, we will need to handle some of your and your baby's personal information. If you need more information about this, you can:

- visit the website: phw.nhs.wales/use-of-site/privacy-notice
- email: PHW.InformationGovernance@wales.nhs.uk, or
- phone: 029 2010 4307

We also keep personal details to make sure that the standard of our service is as high as possible. This includes checking your baby's records if your baby is found to have a condition after having a screening test which showed a 'not suspected' result.

We only ever publish information as statistics and we never publish personal details. We pass on your personal information to health professionals or organisations that need it, including your GP, health visitor and consultant paediatrician. These professionals must protect the personal information in the same way as we do.

All our paper and computer records are stored and processed securely, and the public cannot access them.

Information courtesy of Public Health England

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