Newborn blood spot screening programme

Your baby’s screening result

Phenylketonuria (PKU) is confirmed
Who is this leaflet for?

Your baby's specialist metabolic team has confirmed a diagnosis of phenylketonuria (PKU).

This leaflet will help you to understand the condition and its treatment.

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.

If a baby has PKU, one of these amino acids called phenylalanine (fee-nile-al-an-een) does not break down in the usual way and builds up in the blood.

Without early diagnosis and treatment, PKU can lead to long term health problems including learning difficulties.
Symptoms

If the right treatment is followed, babies with PKU are well in early life and do not develop symptoms.

Without early treatment babies can develop damage to the brain, including learning difficulties.

Treatment

Treatment for PKU involves a special low-phenylalanine diet. The aim of the diet is to reduce the build-up of phenylalanine which can cause learning difficulties and brain injury.

As high protein foods and milk (including breast milk and normal infant formula) have to be limited, a special infant formula is given to meet all nutritional requirements. This special infant formula is very important because it allows normal growth and development as well as helping to reduce the build-up of harmful toxins in the brain.

Your specialist metabolic dietitian will teach you how to measure and control the amount of protein you give to your baby, including breast milk feeds.

Your baby will need regular blood tests to monitor phenylalanine levels in their blood.

Babies with PKU benefit significantly from effective treatment and can live healthy and active lives.
Your questions answered

Who can I ask for advice and support?

The paediatric or metabolic clinician responsible for your baby’s care will be happy to discuss any queries you might have.

Will my other children need to be tested?

Your other children might be at risk of PKU even though they might have never shown any symptoms to date. It is therefore very important to get them tested if they have not been previously screened for PKU.
What about future children?

A new baby from the same parents has a 1 in 4 chance of having PKU. It is important that they are tested 48 to 72 hours following birth. You should tell your midwife and GP there is a family history of PKU. You should ask for a referral to a paediatrician or genetic counsellor and make a birth plan taking their advice into account. Make sure the birth plan is written in your notes.

A small number of people carry the gene for PKU. These are known as carriers. If both parents are carriers, the baby has a 1 in 4 chance of having the condition.
More information and support

- National Society for Phenylketonuria (NSPKU)
  Tel: 030 3040 1090 - Helpline
  Email: info@nspku.org
  Web: www.nspku.org

- NHS Newborn Blood Spot Screening Programme: www.nhs.uk/bloodspot
Contact details for your specialist metabolic team:

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<td>Consultant</td>
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Find out how Public Health England and the NHS use and protect your screening information at www.gov.uk/phe/screening-data.