



# Newborn bloodspot screening programme

## Your baby's screening result

### Homocystinuria (HCU) is suspected

#### Your baby's screening result

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

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

#### What is HCU?

Homocystinuria (HCU), pronounced ho-mo-sis-tin-ur-ee-a, is a rare but treatable inherited disorder that prevents the normal breakdown of protein.

Babies with HCU inherit two faulty copies of the gene for HCU, 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 HCU do not have one of the enzymes that help break down one of the amino acids. This causes a harmful substance called homocysteine to build up in their blood and urine.

Babies with HCU benefit significantly from early treatment and can live healthy and active lives.

Without early diagnosis and treatment, babies with HCU can develop long-term health problems, including learning difficulties.

## Treatment

The level of homocysteine may be controlled by giving vitamin B6 (pyridoxine). If this does not work, HCU can be treated with a special low-protein diet and dietary supplements. Your baby may also be given medication.

These treatments prevent a build-up of homocysteine in the blood. If your child has HCU, a specialist metabolic team will see them regularly.

## 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 HCU.

If HCU is confirmed, the specialist metabolic team will give your baby a trial of pyridoxine for a week to see if this reduces homocysteine to a safe level.

If the level remains high, your specialist metabolic team will:

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

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

## More information and support

- Metabolic Support UK (The National Information Centre for Metabolic Diseases) provides information and support for people with HCU and their families:  
[www.metabolicsupportuk.org](http://www.metabolicsupportuk.org)
- Newborn Bloodspot Screening Wales:  
[phw.nhs.wales/newborn-bloodspot-screening](http://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)	



## 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](http://phw.nhs.wales/use-of-site/privacy-notice)
- email [PHW.InformationGovernance@wales.nhs.uk](mailto: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.



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