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

Maple syrup urine disease (MSUD) is suspected

Your baby's screening result

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

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

What is MSUD?

Maple syrup urine disease (MSUD) is a rare but treatable inherited disorder that prevents the normal breakdown of protein.

Babies with MSUD inherit two faulty copies of the gene for MSUD, 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 MSUD do not have one of the enzymes that help break down some of the amino acids. This causes harmful substances to build up in their blood and urine. This can leave a sweet smell in their urine.

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

Without early diagnosis and treatment they can develop serious illness and damage to the brain.

Treatment

MSUD can be treated with a special low-protein diet and dietary supplements. This prevents the build-up of the harmful substances. Your baby may also be given medication.

It is important that your baby is fed regularly and does not go for long periods without eating. They also need to see their specialist metabolic team regularly.

If your baby is unwell in any way it is important to follow medical advice. Babies that become very unwell might need intensive-care treatment including dialysis (where their blood is filtered to remove harmful substances).

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 MSUD.

If MSUD is confirmed, your specialist metabolic team will:

- explain how to give your baby the special low-protein diet (if your baby is breastfed, you can continue breastfeeding)
- give you any special dietary supplements and medications your baby will need
- explain how to use an emergency feed during illness
- explain what to do if your baby is not feeding well
- give you written information about MSUD 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.

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

Your questions answered

How will I know if my baby is ill and what should I do?

Babies who have MSUD can become poorly within the first few days of life. They might vomit, be sleepy, develop breathing difficulties, have abnormal movements (spasms that cause the head, neck and spine to arch backwards), and become cold. You might also notice a sweet smell in their urine when you change their nappy.

You should not ignore these symptoms. If left untreated, babies with MSUD can have fits and slip into a coma, which can be life-threatening.

If you are worried that your baby is ill, contact a member of your specialist metabolic team.

If you cannot contact your specialist metabolic team you should take your baby to your local accident and emergency department as soon as possible.

Take any information that you have been given about MSUD to the hospital with you.

More information and support

- Metabolic Support UK (The National Information Centre for Metabolic Diseases) provides information and support for people with MSUD and their families:
  www.metabolicsupportuk.org

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

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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