

Maple syrup urine disease (MSUD): summary

Public Health England (PHE) created this information on behalf of the NHS. In it, the word 'we' refers to the NHS service that provides screening.

This information is for parents if their baby is suspected of having MSUD or has been diagnosed with MSUD following their newborn blood spot screening test ('heel prick test'). It will help you and your healthcare professionals to talk through the next stages of your baby's care.

About MSUD

Maple syrup urine disease, or MSUD, is a rare but treatable inherited metabolic disorder that prevents the normal breakdown of protein. Babies with MSUD inherit 2 faulty copies of the gene for MSUD, one from each parent.

Babies with MSUD may present with symptoms of feeding problems, sleepiness and abnormal movements even before the newborn screening result is reported. They can go on to have seizures and slip into a coma, which can be life-threatening.

If MSUD is not treated promptly, it can lead to long-term health problems, including brain damage. Newborn screening allows the diagnosis of MSUD to be made early which can improve the long-term outcome.

Screening and diagnosis of MSUD

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 leucine suggests your baby may have MSUD. 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 contacted by the metabolic team and instructed to immediately take your baby to the local hospital for admission, even if they are well.

Many babies with MSUD will already be unwell and may be in hospital before the screening result is reported.

The local hospital will start appropriate treatment for MSUD and arrange transfer to the metabolic specialist centre for further management.

Blood tests will be carried out to confirm if your baby has MSUD, but treatment will be started based on the screening result as the condition can be life-threatening.

Treatment

If your baby does have MSUD, the metabolic team will explain the condition in more detail and answer any questions you might have. They will start your baby on medical treatment and a special diet, and they will arrange regular follow-up appointments. Following the diet and treatment instructions is very important for your baby's health.

What to do if your baby gets ill

If your baby becomes ill, they might have an episode known as a metabolic crisis (also known as metabolic decompensation). A metabolic crisis can lead to serious illness and long-term brain damage, and can be life threatening.

To help prevent this from happening, you will be taught to give an emergency regimen, which involves specialist feeds and frequent feeding. If your baby becomes very unwell, they might need to be admitted to hospital.

The metabolic team will teach you how to look after your child during illness.

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 detailed information on MSUD, see <https://www.gov.uk/government/publications/msud-confirmed-diagnosis-description-inbrief>.

Further information can be found from Metabolic Support UK 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 British Inherited Metabolic Diseases Group (BIMDG) website has a TEMPLE booklet about MSUD at <http://www.bimdg.org.uk/site/temple.asp> and emergency regiment guidelines at <https://bimdg.org.uk/site/guidelines.asp>.

NHS.UK has information about MSUD at <https://www.nhs.uk/conditions/maple-syrupurine-disease/> and about newborn blood spot screening at <https://www.nhs.uk/conditions/baby/newborn-screening/blood-spot-test/>.

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