Your baby's screening result: Maple syrup urine disease (MSUD) suspected

Your baby's newborn bloodspot test (heel prick) suggests they may have maple syrup urine disease (MSUD).

This does not confirm MSUD – more tests are needed.

What is MSUD?

Maple syrup urine disease (MSUD) is a rare but treatable inherited metabolic disorder.

Babies with MSUD do not have enough of the enzyme needed to break down part of the protein found in food. Without this enzyme, certain substances can build up in the body. If left untreated, it can lead to serious illness and long term health problems.

With early diagnosis and treatment, babies with MSUD can grow and develop normally, and people with MSUD can live healthy, active lives.

What happens next?

A specialist metabolic team will arrange additional tests to confirm whether your baby has MSUD.

It may take a few days for these test results to be reported.

If your baby becomes unwell or you are concerned about poor feeding, please seek medical attention.

If tests confirm your baby does have MSUD, you will receive further support and care from the metabolic team who will explain the condition in more detail, answer your questions and start treatment promptly.

How you might be feeling:
It's completely natural to feel worried or unsure when your baby needs further testing. You can find more information and support from the resources below:

Metabolic Support UK

- Website: www.metabolicsupportuk.org
- Phone: 0845 241 2173 or 0800 652 3181
- Email: contact@metabolicsupportuk.org

The British Inherited Metabolic Diseases Group (BIMDG) website has a useful booklet about MSUD

Contact details for the metabolic team:

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Clarity approved by Plain English Campaign