# MCADD: an overview

This information is for parents if their baby is suspected of having MCADD or has been diagnosed with MCADD 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 MCADD

Medium-chain acyl-CoA dehydrogenase deficiency, or MCADD, is a rare but treatable inherited metabolic disorder that prevents the normal breakdown of fat. Babies with MCADD inherit 2 faulty copies of the gene for MCADD, one from each parent.

MCADD becomes a problem during prolonged fasting and illness because fat cannot be broken down quickly enough and harmful substances build up. Untreated MCADD can cause serious illness, and can be life threatening, but with newborn screening and early treatment this can be prevented.

# Screening and diagnosis of MCADD

## 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 a substance called octanoylcarnitine (C8) in the blood. A high level of octanoylcarnitine (C8) suggests your baby may have MCADD. This is called a screen positive result.

## Diagnostic tests

If your baby has a screen positive result, you will be seen by a metabolic doctor, dietitian and nurse specialist (the ‘metabolic team’). The team will provide advice and support. Blood and urine tests will be carried out to confirm if your baby has MCADD.

You will need to wait a few days for the test results to be reported. During this time, you can continue to breastfeed or give normal infant formula. You should feed your baby every 3 to 4 hours, day and night, or more often if demanded. If your baby becomes unwell, you should take them to hospital for further assessment.

# Treatment

If your baby does have MCADD, the metabolic team will explain the condition in more detail and answer any questions you might have. They will teach you how to look after your child during illness, and they will arrange regular follow-up appointments. Following their advice on illness management 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.

# More information and support

The metabolic team will be happy to discuss any queries you might have.

For further information, see MCADD: further information at https://www.gov.uk/government/publications/mcadd-description-in-brief.

Further information can be found at Metabolic Support UK at www.metabolicsupportuk.org.

The Metabolic Support UK team can be contacted at:

- 0845 241 2173 or 0800 652 3181
- contact@metabolicsupportuk.org

The British Inherited Metabolic Diseases Group (BIMDG) website has the emergency regimen guidelines at bimdg.org.uk/site/guidelines.asp and a TEMPLE booklet about MCADD at bimdg.org.uk/site/temple.asp.

NHS.UK has information about MCADD at www.nhs.uk/conditions/mcadd/ and newborn blood spot screening at www.nhs.uk/conditions/baby/newborn-screening/blood-spot-test/.



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