MCADD: detailed information

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 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. Babies with MCADD inherit 2 faulty copies of the gene for MCADD, one from each parent.

Babies with MCADD have a deficiency of the enzyme called medium-chain acyl-coA dehydrogenase, which is needed to turn fat into energy. Fat is the body’s main energy store and is used for energy supply during fasting. The longer you fast for, the more you use fat to produce energy.

This deficiency becomes a problem during prolonged fasting and illness because fat cannot be broken down quickly enough and harmful substances build up. These substances can lead to serious symptoms, such as excessive drowsiness and coma, which can be life threatening. However, 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 detail and answer any questions you might have
- arrange regular follow-up appointments
- provide you with contact details for the metabolic team and written information

The information the team gives will cover:

- safe fasting times for day to day management (this changes with age – older children can fast for much longer than babies)
- a special feed to use during illnesses (called the emergency regimen)

The metabolic team will see you regularly throughout your child's life and your child will remain under their care.

Feeding your baby

Babies with MCADD should feed regularly (every 3 to 4 hours day and night) and should not fast for more than 6 hours. You can give them breastmilk or standard infant formula.

As your baby grows, they will be able to fast for longer periods. They should eat a normal healthy diet, follow a healthy lifestyle and be treated like any other child.
Very occasionally, babies are prescribed a specialised infant formula for another medical problem. If your baby is prescribed a specialised infant formula, you should check with your metabolic dietitian whether it is suitable. Your baby should not be given a specialised feed which has added medium-chain fats.

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). This is caused by a rapid build-up of harmful substances which can make them severely unwell.

Symptoms of a metabolic crisis (metabolic decompensation) include:

- irritability
- sleepiness and non-responsiveness
- floppiness and falling over
- poor feeding
- breathing difficulties
- seizures
- coma

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.

The emergency regimen

The emergency regimen is a glucose polymer feed. It is given regularly, day and night, to provide energy and help limit the breakdown of body fat.

You should start the emergency regimen if your baby becomes unwell or is not tolerating their usual feeds. Your metabolic dietitian will teach you:

- how to prepare the emergency feed
- how much feed to give your baby
- how often you should feed your baby

Your GP will be asked to prescribe the glucose polymer powder.

Find emergency regimen guidelines at the British Inherited Metabolic Diseases Group (BIMDG) at bimdg.org.uk/site/guidelines.asp.

You should contact the metabolic team at the start of any illness to let them know your baby is unwell and that you have started the emergency regimen.

Signs of illness may include:

- high temperature
- vomiting
- cough/cold
- sore throat
- chest infection
- diarrhoea
- not being their usual self

You should take your baby to hospital immediately for further assessment if they do not tolerate the emergency regimen or their symptoms are getting worse.

You should take with you any information you have been given about MCADD, including your:

- emergency regimen instructions
- BIMDG guidelines
- glucose polymer powder and scoops

Long-term outlook

With prompt use of the emergency regimen and avoidance of prolonged fasting, the outcome is usually very good and most children will avoid any long-term health problems.

Your other children

At-risk siblings

Children from the same parents have a 1 in 4 chance of having MCADD. Your other children might be at risk of MCADD even if they have never shown any symptoms. It is therefore very important to get them tested if they have not been previously screened for MCADD. Your metabolic team will be able to arrange this testing.

Future children

A new baby from the same parents will also have a 1 in 4 chance of having MCADD. When you find out that you are pregnant, you should tell your midwife and GP that there is a family history of MCADD. You should also inform your metabolic team early in the pregnancy.

The metabolic team will write a birth plan for you. This will include advice on an early screening test for your new baby which should be taken between 24 and 48 hours after birth. This blood test will be in addition to the routine newborn blood spot screening test.

The birth plan will be given to you and shared with your obstetrician and local midwifery team. You should notify your metabolic team once you have given birth so they can ensure the correct blood tests are taken and sent to the screening laboratory without delay.

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.

Further information can be found at Metabolic Support UK at www.metabolicsupport.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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