Newborn blood spot screening programme

Your baby’s screening result

Medium-chain acyl-CoA dehydrogenase deficiency (MCADD) is confirmed
Who is this leaflet for?

Your baby’s specialist metabolic team has confirmed a diagnosis of medium-chain acyl-CoA dehydrogenase deficiency (MCADD).

This leaflet will help you to understand the condition and its treatment.

What is MCADD?

MCADD is a rare but treatable inherited disorder.

Babies with MCADD have inherited two faulty copies of the gene for MCADD, one from each parent.

Babies with MCADD have a problem breaking down fats quickly enough to produce energy.

MCADD only causes problems when fats need to be broken down quickly – for example if a baby has not eaten for a long period or if they have an infection. If this happens it may cause low blood sugar and a build-up of fats. This can create toxic substances that can lead to serious symptoms.

Babies with MCADD benefit significantly from effective treatment and can live healthy and active lives.

Without early treatment, the condition can lead to serious illness and possibly death.
Symptoms of MCADD

Babies with MCADD are at risk of developing the following symptoms:

- poor feeding
- drowsiness
- sleepiness
- vomiting
- lethargy
- seizures

These symptoms can be controlled with treatment. Left untreated, babies with MCADD can deteriorate, have fits and slip into a coma which can be life threatening.

When babies with MCADD become ill, they might show symptoms of metabolic crisis. A metabolic crisis is a period of time when the effects of the condition make your child seriously ill. Some babies with MCADD might not develop symptoms of a metabolic crisis until later on in the first year of life or later in childhood. Causes of a metabolic crisis can include an infection such as a stomach upset or vomiting illness.
Treatment

Babies with MCADD do not need any special medications. Day to day management is to avoid prolonged periods without eating.

Babies with MCADD should feed regularly and it is fine for them to be breast or bottle fed. As they grow, they should eat a normal healthy diet and be treated like any other child.

The specialist metabolic team will see you regularly throughout your child’s life and your child will remain under their care. The specialist dietitian will discuss age appropriate advice on feeding. For example they will discuss when to wean, safe fasting times and illness management. This includes the preparation and use of emergency regimen feeds.
If your baby is ill

During illness, babies with MCADD need to be managed correctly to prevent serious illness or even death.

If your baby becomes ill or is not feeding well they should be given a special high sugar drink (also known as glucose polymer). This is called the emergency regimen. It is given without delay and frequently, day and night. Sugar free and low calorie drinks are not suitable.

The emergency regimen is given to provide the body with plenty of energy and to help prevent the breakdown of body fats.

The metabolic dietitian will provide detailed instructions on how to give the emergency regimen.

If you are worried that your baby is not improving or not taking the entire emergency regimen then you should take them to the accident and emergency department of your local hospital and contact your metabolic team.
Your questions answered

What if my baby vomits the emergency regimen drinks?

If your baby cannot keep down their emergency feeds, continues to vomit or has repeated episodes of diarrhoea despite using the emergency feeds, you should take them to hospital immediately.

You should also take your baby to hospital immediately if they seem unusually sleepy, irritable or have rapid breathing.

What if my baby needs to go into hospital?

If you have to take your baby into hospital it is important to take your British Inherited Metabolic Disease Group (BIMDG) accident and emergency guide with you. You should also take your MCADD dietary management guidelines and glucose polymer powder.

These emergency guidelines are also available on the BIMDG website at www.bimdg.org.uk.

It is also helpful to contact your specialist metabolic team to say you are on your way but do not delay going to hospital.

What if I lose my documents?

Contact your specialist metabolic team as they can send new copies.

Should my other children be tested?

Your other children might be at risk of MCADD even if they have never shown any symptoms to date. It is therefore very important to get them tested if they have not been previously screened for MCADD.
Your questions answered

What about future children?
A new baby from the same parents has a 1 in 4 chance of having MCADD. It is important that they are tested 24 to 48 hours following birth.

You should tell your midwife and GP there is a family history of MCADD. You should ask for a referral to a paediatrician or genetic counsellor and make a birth plan taking their advice into account. Make sure the birth plan is written in your notes.

A small number of people carry the gene for MCADD. These are known as carriers. If both parents are carriers, the baby has a 1 in 4 chance of having the condition.
Who can I ask for advice and support?

The paediatric or metabolic clinician responsible for your baby’s care will be happy to discuss any queries you might have.

More information and support

- CLIMB (The National Information Centre for Metabolic Diseases) provides information and support for people with MCADD and their families: www.climb.org.uk

- NHS Newborn Blood Spot Screening Programme: www.nhs.uk/bloodspot
Contact details for your specialist metabolic team:

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<td>Metabolic dietitian</td>
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Notes

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<td>March 2019</td>
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More information: [www.nhs.uk/bloodspot](http://www.nhs.uk/bloodspot)

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