Newborn blood spot screening programme

Your baby’s screening result

Medium-chain acyl-CoA dehydrogenase deficiency (MCADD) is suspected
Your baby’s screening result

The result of your baby’s ‘heel prick’ screening blood test suggests they might have medium-chain acyl-CoA dehydrogenase deficiency (MCADD). A specialist team will do further tests to confirm this result.

This leaflet gives some information about MCADD and explains what happens next.
What is MCADD?

Medium-chain acyl-CoA dehydrogenase deficiency (MCADD) is a rare but treatable inherited disorder.

Babies with MCADD inherit 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 when a baby has not eaten for a long period or when they are unwell. If this happens, MCADD can cause low blood sugar and a build-up of certain fats. This can make toxic substances that can lead to serious symptoms.

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

Without early diagnosis and treatment, MCADD can lead to serious illness and possibly death.
**Treatment**

Babies with MCADD should feed regularly, eat a healthy diet and be treated like any other child. They do not need special medications. They do not need a special diet but should avoid long periods without feeding.

Children with MCADD will be seen regularly by a specialist metabolic team.

If your baby is unwell in any way it is important to follow medical advice. When they are unwell they may need to go into hospital for treatment. Take any information you have about MCADD with you.
What happens next?

You have been given an appointment with a specialist metabolic team who will:

- discuss the screening test result with you
- arrange blood and urine tests for your baby
- explain how these tests can confirm if your baby has MCADD

If MCADD is confirmed, the specialist metabolic team will:

- explain how to use an emergency feed during illness
- explain what to do if your baby is not feeding well
- let your GP know about your baby’s tests and MCADD
- give you written information about MCADD for you to share with your family, GP and local hospital
- answer any questions you may have
- arrange a follow-up appointment to discuss the test results

Until you see the specialist team, you can breast feed or bottle feed your baby with normal infant formula. You should feed your baby every 3 to 4 hours, day and night, or more often if demanded. If you are concerned about poor feeding or if your baby is unwell, contact your specialist metabolic team.
How will I know if my baby is ill and what should I do?

Babies who have MCADD can become ill within the first few days of life. If they become ill they may:

- feed poorly
- vomit or have diarrhoea
- become drowsy, irritable or not respond normally

You should not ignore these symptoms. If left untreated, babies with MCADD can have fits and slip into a coma which can be life-threatening.

If you are worried that your baby is ill, contact a member of your specialist metabolic team. If you cannot contact your specialist metabolic team you should take your baby to your local accident and emergency department as soon as possible.

Take any information that you have been given about MCADD to the hospital with you.

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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