

Medium Chain acyl CoA Dehydrogenase Deficiency (MCADD) is suspected



5 year old boy
with MCADD

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All records kept relating to newborn blood spot screening meet the requirements of the 1998 Data Protection Act.

This leaflet is based on high-quality research evidence and the views of parents and health professionals. It has been produced by the UK Newborn Screening Programme Centre, which is funded by the Department of Health for the whole of the UK.



Photography: Children's photographs were provided with kind permission from their parents who are on the UKNSPC MCADD Board. Picture of the baby being screened taken by Lois Cavanagh.

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What is my baby's screening result?

When your baby was about one week old, your midwife took some blood from your baby's heel. This blood test is offered to all newborn babies and tests for some rare conditions, including MCADD.

The screening test result suggests that your baby may have MCADD. Your baby now needs further tests to check whether she/he has MCADD or not.

This leaflet gives you some information about MCADD and what happens next.

What is MCADD?

During long periods between eating, the body breaks down its own fat stores to produce energy. People with MCADD lack one of the enzymes needed to do this. They can break down the stored fat partly but not completely. There is a hold up at the 'medium chain fat' step where the enzyme needed to complete the breakdown is not working properly. This causes a build up of medium chain fats.

Sometimes we need to break down fats quickly, for example, when we have not eaten for some while or when we have an infection. People with MCADD can't do this. The medium chain fats can build up and make toxic substances that may lead to serious symptoms.

If this condition is not diagnosed early or is ignored and not treated by following simple advice from a specialist medical team, it could lead to serious illness and possibly death. Fortunately - once diagnosed - MCADD is usually quite straightforward to manage and children with this condition usually lead healthy normal lives.



5 month old girl
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Baby being
screened

If your baby becomes unwell before your hospital appointment with the specialist medical team you should seek medical advice. It is important that the healthcare professional you contact is aware of this screening result for MCADD and we suggest that you show them this leaflet.

Where can I find more information or support?

You can find more information about MCADD on the UK Newborn Screening Centre website (www.newbornbloodspot.screening.nhs.uk/resources/index.htm). Please follow the link to Parent Information.

Climb (the National Information Centre for Metabolic Diseases) provides information and support for people with MCADD and their families. You can contact them as follows:

Climb Building,
176 Nantwich Rd,
Crewe CW2 6BG

Telephone helpline: 0800 652 3181 (freephone) or
0845 241 2172

Website: www.climb.org.uk

Email: info.svcs@climb.org.uk



What treatment is available for MCADD?

MCADD is treated by diet. When the child is well, there is no specific dietary management apart from avoiding long periods without food. The well child can be on a normal, healthy diet. During illness an emergency diet of very frequent drinks containing glucose polymer (a type of sugar) is needed.

What happens next?

You have been given an appointment to see a specialist doctor and dietitian. They will

- discuss the screening test result with you
- arrange for your baby to have a blood test and a urine test
- give you more advice about feeding your baby until the results of these further tests are known
- support you now and in the future if your baby has MCADD
- give you advice on what to do if your baby is not feeding well for any reason
- let your family doctor know about your baby's tests and MCADD
- give you written information about MCADD to share with your family and local hospital

Parents of babies with MCADD often ask the following questions:

How should I feed my baby until I see the specialist medical team?

You can breast feed or bottle feed your baby with normal infant formula. You should feed your baby 3 to 4 hourly day and night, or more often if demanded. Your baby should not fast (go without a feed) for longer than **6 hours**. If your baby is not feeding well, contact your doctor. It is important to tell them that MCADD is suspected.

Why do some children have MCADD?

MCADD is an inherited condition due to an altered gene. It is not caused by anything which happened during pregnancy. Everyone carries a few genes that have become altered in some way and are passed on sometimes unknowingly in families. A baby with MCADD has inherited two copies of an altered gene, one from each parent, which together cause MCADD. When two people with this gene alteration have a baby, they have a 1 in 4 (25%) chance in every pregnancy of having a child with MCADD. There is nothing the parents could have done to prevent their child having MCADD.

What is life like for children with MCADD?

Children with MCADD are able to live full and active lives, just like any other children, provided they eat regularly and avoid long periods without food. In case of illness, when a child is not able to eat or keep food down, a visit to hospital may be needed to make sure the child gets glucose quickly.



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