Medium-Chain Acyl-CoA Dehydrogenase (MCAD) deficiency and your baby

You have just learned that your baby had a positive newborn screening result that suggests that baby may have Medium-Chain Acyl-CoA Dehydrogenase (MCAD) deficiency. The information in this leaflet will help you understand more about this condition. You should be seen by a paediatrician within 24 hours of receiving this result and will have an opportunity to ask more questions. We suggest you write these down and bring them to the appointment.

MCAD deficiency screening test

When your baby was about 48 hours old, a blood sample was collected from your baby's heel onto a blood spot (Guthrie) card. The blood was used to test for some rare disorders, including MCAD deficiency. Your baby's screening test result suggests that your baby is very likely to have MCAD deficiency. Further tests are needed to confirm this. Each year in New Zealand about four babies are diagnosed with MCAD deficiency.

MCAD deficiency

MCAD deficiency is what is known as a Fatty Acid Oxidation Disorder. Fat is an important fuel for the body and is stored in our fat cells to be used in between meals. There are many different enzymes that help break down fat. They are required to turn fat into energy and are especially important when we are not eating regularly or when we are unwell. One of these enzymes is called medium chain acyl-CoA dehydrogenase. MCAD deficiency occurs when the enzyme is either missing or not working properly.

Once diagnosed MCAD deficiency is easy to manage with:

- regular feeding of normal food (eg breast milk)
- an emergency management plan for illness.

This is especially important in the first few years of life.

We know that people with this condition, diagnosed through newborn screening and managed with these simple strategies live a normal life.

What will happen next?

Your midwife will talk with you about arranging an appointment for you and your baby, at your local hospital, to see a doctor and their team which may include a nurse and a dietitian.
If you are living in Auckland you will see the National Metabolic Team (a specialist team who look after children with MCAD deficiency). If you are living outside of Auckland you will see a paediatrician and then the National Metabolic Team when they next do a clinic in your area.

The paediatrician will then discuss the next steps with you. This will include:

- discussing the screening test result
- arranging for your baby to have confirmatory blood and urine tests

- giving you advice about feeding your baby until the results of these further tests are known
- giving you advice on what to do if your baby is not feeding well for any reason
- letting your family doctor know about your baby's tests
- giving you more written information about MCAD deficiency to share with your family and if you seek medical care
- answering any questions you have.

Why are babies screened for MCAD deficiency?

One aim of newborn screening is to identify babies who are more likely to have MCAD deficiency so that treatment can be started early. Babies with MCAD deficiency do not seem obviously different from other babies and so without newborn screening, people with MCAD deficiency usually present when their body runs out of fuel during an illness or after a period of fasting. Eventually the blood sugar becomes low and there is not enough energy for the brain, which is very dangerous, and if left untreated can lead to death. This is something that will not happen to your baby if he or she is fed regularly and illness is managed well.

What treatment is available for MCAD deficiency?

When your baby is well, MCAD deficiency is treated with regular breast feeding or infant formula. As your baby gets older, eats solids and participates in family meals, the time between meals can increase and they can have a normal, healthy diet. During illness frequent feeding or an emergency drink containing glucose will be prescribed by your doctor and dietitian and they will give you a plan for when to take your child to hospital and how that should be managed.

How should I feed my baby until I see a paediatrician or get the follow-up test results?

You should continue to feed your baby as you are currently doing, either breast feed or bottle feed with standard infant formula, every three to four hours day and night, or more often if baby is hungry and wanting to be fed. Your baby should not fast (go without food) for longer than six hours.

If your baby is not feeding well, or becomes sick, or you are in any way concerned about your baby contact your midwife or in an emergency, go to the emergency department at your hospital. It is important to tell them that MCAD deficiency is suspected.

What is life like for people with MCAD deficiency?

Children with MCAD deficiency diagnosed on newborn screening live full and active lives, just like any other children, provided they eat regularly and avoid long periods without food. If your child becomes unwell, is not able to eat their usual foods and is not tolerating their emergency drink they will need to go into hospital.

Why do some children have MCAD deficiency?

MCAD deficiency is a genetic condition that has been passed down from both parents. It is no one's fault that it has happened and it is not caused by anything that happened during pregnancy.

Where can I find more information or support?

Talk with your paediatrician or midwife.

View the following websites:

- www.nsu.govt.nz (for information on the Newborn Metabolic Screening Programme).
- http://newbornbloodspot.screening.nhs.uk/mcaddparentinfo
- www.fodsupport.org/mcad-fam.htm

MINISTRY OF HEALTH
MANATŪ HAUORA
National Screening Unit
New Zealand Government
www.nsu.govt.nz

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