



## *fact sheet for parents*

# Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency (MCAD)

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### **How common is MCAD deficiency?**

Metabolic disorders are quite rare. MCAD deficiency is one of the more common metabolic disorders. It has been found that up to 1 in every 40 healthy people carry one gene for MCAD deficiency in their body (These people are called CARRIERS). MCAD deficiency is passed on from parents to child in 'an autosomal recessive manner'.

Two healthy parents, if both carry the gene for MCAD deficiency, have a 1 in 4 chance of having a child with this condition. The child would be at risk of inheriting two MCAD genes, one from each parent and the child would then be born with MCAD deficiency. MCAD deficiency will affect 1 in 18,500 children. In South Australia, we would expect to see 1 child diagnosed with this condition each year. (In New South Wales 5-6 children and 3-4 children in Victoria).

### **What is MCAD deficiency?**

Medium-chain acyl-Coenzyme A dehydrogenase (MCAD) is a special enzyme, a type of protein, which is part of a chain of enzymes which act together in the body to turn fat into energy.

MCAD deficiency is the commonest of a

group of disorders which affect how the body can break down stored fat. This group of conditions is known as 'fatty acid oxidation defects' and has only recently been recognised. MCAD deficiency was first described in the medical journals at the end of 1982, but it was only in 1990 that the genetic mistake in most cases of MCAD deficiency was found. Because this is truly a new disorder, many people, including doctors, have not heard of it.

### **Newborn Screening for MCAD Deficiency**

New, "expanded" Newborn Screening Programs using Tandem Mass Spectrometry can diagnose MCAD deficiency in babies 48 hours after birth, before they get sick. Such programs are, in 2005, available in most parts of Australia.

### **Special points of interest:**

- *Babies can safely fast for 6 hours when well but only 4 hours if they have a cold or fever*
- *1 year old children can fast for 8–10 hours when well but only 6 hours if ill*
- *1 in 40 Australians are healthy carriers of MCAD deficiency*
- *Frequent snacks are a good idea if you have MCAD deficiency*

## Why does MCAD deficiency cause problems?

Children have much higher energy needs than adults, mostly because they are growing all of the time. Usually, after a meal, to provide the energy to keep going, our body first uses up the sugar which was contained in the food. In children, this generally lasts for about the first 4 hours, then the body goes on to use stored sugar from the liver, which is called glycogen. If no more food is eaten, the next step involves using stored fat. After about 8 hours of fasting, (not eating or drinking anything other than water) young children start to use ketones, which are the fuels which come from the break down of fat, as an energy source. (This is very different from adults, who only start to use stored fat after about 24 hours of fasting).

Because children with MCAD deficiency cannot make enough ketones, they rely on glucose, which then gets used up. This is why children with MCAD deficiency are prone to low blood glucose, (hypoglycaemia) if they fast for any length of time. Sometimes the low blood sugar causes fits or the child may go into a coma. The coma is partly due to a build-up of 'medium-chain fatty acids' which cannot be broken down, and also partly due to low blood sugar.

However, children with MCAD deficiency are perfectly well so long as they are able to eat often and do not get to the stage where the body needs to use these breakdown products of fat. Some children with MCAD deficiency have never been sick and are only found after a brother or sister has been diagnosed.

The major problem for children with MCAD deficiency is having a virus, with fever, vomiting and diarrhoea, or a sore throat which makes small children not keen to eat or drink. Having a virus means the body needs more energy to fight off the virus and children need to eat more often especially if there is a fever. A good rule of thumb is that children with MCAD deficiency, once they are over about 6 months old, need to eat or drink at least every 10 hours if they are well, or at least every 6 hours if they are sick. If your child fasts for longer than this, or vomits, or is unusually drowsy, they are at risk of getting seriously sick and should be seen promptly by someone who understands MCAD deficiency, such as the specialist who looks after your child. The doctors at the Casualty Departments of major children's hospitals should also be familiar with MCAD deficiency. If in doubt, it is best to ask.

## What is Carnitine deficiency?

Some children (and adults) with MCAD deficiency also have carnitine deficiency.

Carnitine (*3-hydroxy, 4-N-trimethylaminobutyric*

*acid*) is in all cells. It comes from red meat and dairy products as well as being made in the body from the amino acids lysine and methionine. Within the body, carnitine is almost all in the skeletal and cardiac muscle. The main role of carnitine is transport of fatty acids into mitochondria, the cells' energy factories. Carnitine deficiency occurs because carnitine "mops up" excess medium chain fatty acids, making octanoyl carnitine. If it happens, carnitine deficiency is easily treated with carnitine mixture or tablets, usually three times a day.

## Fever

Some new research by Dr Nils Gregersen from Denmark suggests that the MCAD enzyme is less stable (breaks down more quickly) at higher temperatures. We now recommend treating fevers in children with MCAD using medicines such as paracetamol (Acetaminophen) or Ibuprofen.

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