



## MCAD deficiency

(Medium chain acyl coenzyme A dehydrogenase deficiency)

### A fact sheet for parents/carers

#### What is MCAD deficiency?

**MCAD deficiency** is a genetic disorder caused by the deficiency for an enzyme which breaks down fats to give us energy. An **enzyme** is a protein made in the body, which helps a chemical reaction to proceed. Each of the chemical reactions, or steps, in the bodies' metabolism needs a specific enzyme to function.

MCAD is part of a chain of enzymes which act together to break down fats into energy. Babies and children with MCAD deficiency are at danger of developing illness with low blood sugar during episodes of prolonged fasting.

**Fat stores in the body.** Energy is stored in the body either as a glucose complex (glycogen) or as fat. When we need energy we can use up the glucose stores, after which we need to mobilise the fat stores. These are broken down into fatty acids, which are themselves broken down into shorter lengths ("chain lengths"), each shortening producing energy. Children with MCAD deficiency can break down fat to some extent, but cannot do this very fast, as there is a hold-up at the medium chain length step. If a child tries to break down fats fast, the banked up medium chain fats form toxic substances. Giving glucose will immediately switch off the breakdown of fats.

#### How common is MCAD deficiency?

MCAD deficiency is one of a group of rare disorders which affect the body's capacity to

break down stored fats at times when other energy sources are unavailable. 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 literature at the end of 1982.

Because this is only a recently described disorder, many people, including health professionals, have not heard of it, although it is the most frequently diagnosed of the fatty acid oxidation disorders.

MCAD studies in the family suggest that between around 1 in 80 healthy people are carriers of MCAD deficiency. MCAD deficiency is inherited in what we describe as an autosomal recessive manner. This means that two healthy parents, if both are carriers for MCAD deficiency, have a 1 in 4 chance of having a child with this condition with each pregnancy. We would therefore expect MCAD deficiency to affect approximately one child in every 25,000. Far fewer than this are diagnosed, and it seems likely that perhaps two thirds of babies with MCAD deficiency will never have any symptoms. Unfortunately, it is not possible to predict which babies or children will develop symptoms, and as the problems can be very serious and even fatal, it is vital to prevent these.

#### When does MCAD deficiency cause problems?

Children have much higher energy requirements than adults, mostly because they are continually growing. Usually, after

a meal, to provide the energy to keep us going, our body first uses up the sugar (glucose) which was contained in the meal. This generally lasts for about the first 4 hours, then the body goes on to use stored glucose from the liver, which is called glycogen. The next step involves using stored fat, which is the body's way of storing excess energy from meals. After about 8 hours of fasting, (not eating or drinking anything but water), young children start to break down fat to form ketones, a metabolic fuel, as an energy source. This is very different from adults, who only start to use fat after about 24 hours of fasting.

Because children with MCAD deficiency cannot make ketones fast enough to provide an alternative source of energy, they rely on glucose, which then gets used up. This is why children with MCAD deficiency usually have 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 substances known as medium chain fatty acids, which can't be broken down, and partly due to low blood sugar. They can get sick before the blood sugar is really low.



However, children with MCAD deficiency are perfectly well so long as they are able to eat regularly 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 to have the condition after a brother or sister has been diagnosed

The major problem for children with MCAD deficiency is when they have a viral illness particularly if there is vomiting and diarrhoea, or a sore throat, which makes small children reluctant to eat or drink. Having a virus means the body requires increased energy to fight off the virus, and children need to eat more often under these conditions, particularly if there is a high temperature. A good rule of thumb is that babies with MCAD deficiency need to feed every 4 hours during the day and every 5-6 hours overnight. By 6 months of age they need to feed regularly during the day and can sleep for no longer than 10 hours overnight. If they are sick, they should be fed every 6 hours at night.

If a child fasts for longer than this, or vomits, or becomes unusually drowsy, the child should be seen promptly by someone who understands the condition, such as the paediatrician or the metabolic doctor who supervises the child's care. The doctors at the casualty departments of the Children's Hospital at Westmead, the Sydney Children's Hospital, Randwick and other big hospitals with paediatric sections should also be familiar with MCAD deficiency. Even so it is important to take this information sheet to the hospital.

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For further information please contact your paediatrician or the NSW Newborn Screening Programme  
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