

Medium chain acyl-CoA dehydrogenase (MCAD) deficiency:

A fact sheet for parents/carers

November 2006

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 deficiency is part of a chain of enzymes which act together to break down fats into energy. Babies and children with MCADD 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 MCADD 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. MCADD was first described in the medical literature at the end of 1982.

It is estimated that approximately 1 in 15,000 children will be identified with MCADD.

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 using 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 disorder 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 six 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 six 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 disorder, such as a paediatrician or the metabolic doctor who supervises the child's care.