

Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)

What is MCAD?

Medium chain acyl-CoA dehydrogenase deficiency (MCAD) is a condition in which the body is unable to break down certain fats. It is considered a fatty acid oxidation condition because people affected with MCAD are unable to change some of the fats they eat into energy the body needs to function. Instead, too many unused fatty acids build up in the body. If untreated, MCAD can cause breathing problems and low blood sugar. However, if the condition is detected early and proper treatment is begun, people affected by MCAD can often lead healthy lives.

What Causes MCAD?

When we eat food, enzymes help break it down. Certain enzymes help break down fats into their building blocks, fatty acids. Fatty acids are built like chains and come in many lengths. They are categorized as either short, medium, long, or very long. Different enzymes work on breaking down different lengths of fatty acids. An enzyme called medium-chain acyl-CoA dehydrogenase is in charge of breaking down medium-length fatty acids.

If your baby has MCAD, your baby's body either does not make enough or makes non-working medium-chain acyl-CoA dehydrogenase enzymes. When this happens, your baby cannot use medium-length fatty acids for energy. This is harmful because your baby's body needs fat for energy when his or her body runs out of sugars (such as between meals). The heart also needs fatty acids for energy.

MCAD is an autosomal recessive genetic condition. This means that a child must inherit two copies of the non-working gene for MCAD, one from each parent, in order to have the condition. The parents of a child with an autosomal recessive condition each carry one copy of the non-working gene, but they typically do not show signs and symptoms of the condition. While having a child with MCAD is rare, when both parents are carriers, they can have more than one child with the condition.

What Symptoms or Problems Occur with MCAD?

[Symptoms are something out of the ordinary that a parent notices.]

MCAD can cause bouts of illness called metabolic crises. Children with MCAD often show symptoms for the first time between 3 months and 2 years of age.

Some of the first signs of a Metabolic Crisis are:

- sleeping longer or more often
- behavior changes (such as crying for no reason)
- irritable mood
- poor appetite

If a metabolic crisis is not treated, a child with MCAD can develop:

- breathing problems
- seizures
- intellectual disability
- cerebral palsy
- coma, sometimes leading to death

What is the Treatment for MCAD?

The following treatments are often used for children with MCAD:

1. Do not go a long time without food – Babies and young children with MCAD need to eat often to avoid low blood sugar or a Metabolic Crisis. They should not go without food for more than 4 to 6 hours. Some babies need to eat even more often. It is important that babies be fed during the night. They need to be woken to eat if they do not wake up on their own. Young children with MCAD may need to have a starchy snack (such as bread, cereal, and rice) before bed and another during the night.

They may need another snack first thing in the morning. Your dietitian can give ideas for good night-time snacks. Dietitians know the correct foods your child can eat. Most teens and adults with MCAD can go without food for up to 12 hours without problems when they are well. They need to continue the other treatments for life.

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2. Diet – Sometimes a low-fat, high carbohydrate diet (such as vegetables, fruits, grains) is advised. Your dietitian can create a food plan with the right type and amount of fat your child needs. Ask your doctor whether or not your child needs to have any changes in his or her diet.

3. L-carnitine – L-carnitine (Carnitor) may be prescribed for some children. This is safe and natural and helps body cells make energy. It also helps the body get rid of harmful wastes.

Things to Remember

Always call your doctor when your child has any of the following:

- poor appetite
- low energy or too much sleepiness
- vomiting
- diarrhea
- an infection
- a fever

People with MCAD need to eat extra starchy foods and drink more fluids during any illness – even if they don't feel hungry – or they could develop low blood sugar or a Metabolic Crisis. Children who are sick often don't want to eat. If they won't or can't eat, they may need to be treated in the hospital to prevent problems.