



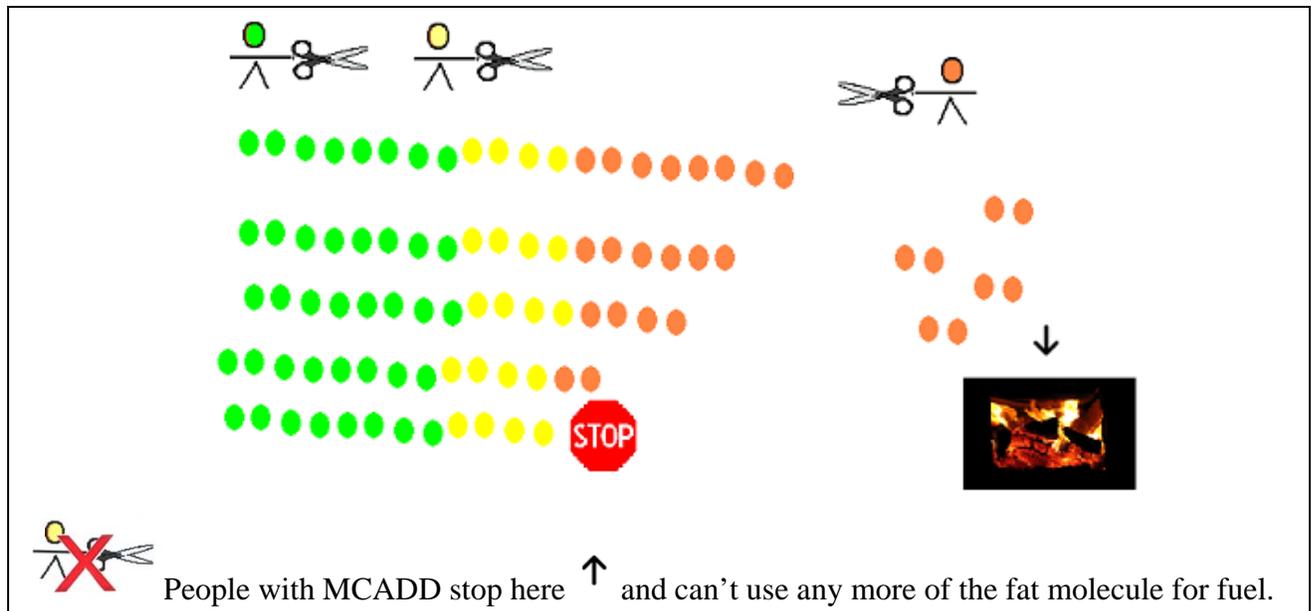
Understanding MCAD Deficiency: (Medium Chain AcylCoA Dehydrogenase Deficiency)

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What is the basic problem? People with MCADD have a genetic problem with chopping up chains of fat to a size small enough to go into the “furnace” (in the mitochondria in cells) and be burned to make energy to run one’s body. It is usually identified by newborn screening, and babies identified that way who follow a special nutrition program have the potential to live healthy normal lives.

The chains of fat we use for fuel to make energy are like strings of beads, and the size they need to be chopped up to is 2 beads long. There are **three kinds of fat “choppers”*** in the mitochondria: (* the real name of any chopper is an “enzyme”)

- Those who chop up the longest pieces (14 beads or more) are called **Long Chain Choppers** 
- Those who chop up the medium-sized pieces (10-12 beads) are called **Medium Chain Choppers** 
- Those who chop up the shortest pieces (8 beads or less) are called **Short Chain Choppers** 



The actual name of a fat chopper of this type is AcylCoA Dehydrogenase.

So, the ones that chop up the medium sized chains are called **Medium-Chain-AcylCoA-Dehydrogenase**, or **MCAD**. If a person does not make enough of this type of chopper, he/she is said to have “MCADD” ... or **Medium Chain AcylCoA Dehydrogenase Deficiency**.

People with MCAD cannot use all the rest of the fat molecule to make fuel, so it is very important that **usable fuel sources like carbohydrate (sugars and starches) should be provided as an alternative fuel**.

Problems with fasting:

During fasting we usually burn fat for fuel, so the person with MCADD may not be able to fast very well. They could run out of fuel and burn the only thing they have available: their blood sugar. That could cause a low blood sugar and the person could be very sick or hurt or he/she could even die. That is why babies with MCADD are not allowed to go more than three hours without being fed ... we don't trust them to be able to burn fat in order to fast well enough to go longer than that. Later they will be able to go longer between feedings, and your baby's doctor will tell you when and how to increase the interval between feedings as baby grows. There are ways to be very sure when the baby can safely fast longer.

Problems if people with MCADD get sick and can't eat:

This person needs to be provided with carbohydrate right away... even Kool-aid or juice or sugar water, or some sugar tucked into baby's cheek are very useful and easy-to-get carbohydrate. Another way to provide a longer-lasting form of carbohydrate is to add raw cornstarch to foods or liquids.

[It does not matter if the food is not usually regarded as a nutritional or traditional food for babies ... the only thing that matters in this emergency situation is safely giving baby something to keep her/his blood sugar from dropping too low.] **Give this sugary or starchy food right away and take the baby to the ER because if baby is very sick he/she may need them to give intravenous glucose. But don't wait until you get there ...give that baby some carbohydrate all the way to the hospital!**

Need for supplemental carnitine:

The person with MCADD will need an **extra amount of a substance that people make called carnitine**. It helps to cart the long fat molecule into the chopping area. [Memory trick: Carnitine = “Cart-It-In”] **People with MCADD need more than they can usually**

make because they also have to haul the medium-chain-length stumps of the fat BACK OUT of the mitochondria. Usually one does not need to empty the trash like this.



This picture of a logging truck illustrates the role of carnitine if you imagine the long trees as long chains of fat. Carnitine “trucks” are needed to get them into the saw mill to cut them up in to small enough bits to go into the furnace.

The MCADD problem affects the ability to chop up the fats AFTER they are carried into the saw mill. MCADD makes one need more carnitine than other people need because they also need to take the unusable stumps of fat back out of the mitochondria so they do not build up.

The baby’s doctor needs to prescribe carnitine, and the amount needs to be increased as the person gets bigger. (It is calculated by body size.) The usual amount for infants with MCADD is in the range of 50-150 mg/kg body weight daily, divided into three doses. This is approximately 25-75 mg per pound the baby weighs. As a rule-of-thumb, some families or home doctors **set up an automatic “bump up” whenever the baby gains another pound.** This works very nicely.

As you can see, the range needed is fairly broad ... some need a lot and some need less. They ALL need SOME. That means that there will likely be adjustments made as we know more about how much an individual baby needs. If the carnitine prescription is set higher than a certain baby needs, he/she may send the extra amount out in the form that smells like a little fish. This is not harmful ... just stinky. All that needs to be done is dial back the prescription in 10% increments until the right amount is provided and the fishy smell goes away.

Carnitine is actually “L-carnitine” and it also comes in forms such as “acetyl-L-carnitine.” The latter form appears to be absorbed more efficiently. Carnitine can be obtained in a liquid form for babies as well as capsules for older people. [Please be sure that the prescription, the pharmacist’s interpretation of the word carnitine, and the substance requested on insurance forms are correct. It is important to check because sometimes people mix up the word carnitine with beta-carotene just because it is a more familiar word. Beta-carotene is the orange pigment in carrots and it a very different substance from carnitine.

Insurance companies sometimes initially refuse coverage for carnitine because it is available “over-the-counter.” However, this use of carnitine is central to the health of a baby with this unusual metabolic condition, so if coverage is refused the doctors should write a letter to make it very clear that the carnitine should be covered in this case. So far, we have always been successful in having it covered for babies with MCADD ... but sometimes we have had to re-submit the request a few times.

Other supplements:

Doctors often prescribe an amino acid called **glycine** (100-200 mg/kg/day) to help some of the fats that are not able to be chopped up adequately be excreted in the urine.

They may also prescribe very generous **vitamin B2 (“riboflavin”)** at 100-200 mg/day for people with MCADD because it may help the affected enzymes work better.

Vitamin D should be provided at 400 iu/day starting right away as recommended by the American Academy of Pediatrics. This can be as a vitamin D drop, or as part of a multivitamin drop. This is important for all babies (not just those with a metabolic disease) for many reasons including decreasing risk of diabetes, cancer, infections and many other serious problems that nobody wants. In our clinic we do check a vitamin D level because many babies here are born with actual vitamin D deficiency. When deficiency is found, we correct the problem with a higher therapeutic dose of vitamin D, after which we have an appropriate follow-up plan to assure that the baby continues to have this critical need met.

A **fish oil supplement** is a good way to assure the availability of two important fats that are very important to the development of the eye and the brain. **Fish oil contains ready-made EPA and DHA**, which are very long chain fats in any case (20 and 22 carbons long) so the fuel available from them is among the best utilized in MCADD. However, this **fish oil supplementation is provided because these fats are used to make critical substances in the body**, so their ability to be used as energy is secondary.

It appears that many individuals have difficulty producing these fats, including the general public, which is why the American Heart Association recommends eating fatty fish twice weekly or taking a fish oil supplement. I prefer to err on the side of caution and assure adequacy and not assume it.

When people are known to have a metabolic disease of any type, they may have more than one system disturbed. In research with patients who have a variety of metabolic diseases, benefit from fish oil supplementation has been found. Supplementation of some of our own patients with metabolic conditions have demonstrated improvement in behavioral measurements at school. What it comes down to is if it were my child, I would give him/her some ready-made fish oil. In fact, I take it myself and make sure all my family members take it too. [And I don’t sell anything and I never will!]

Keep an eye on the total amount of fat and a particular form of fat in food:

Fats in foods naturally come in many sizes. People with MCADD should **avoid eating fats that are naturally high in medium-chain sized fat**. Coconut oil is one that is especially high in these fats, and the person with MCADD cannot use this fat for fuel at all. Medium chain fats are called **MCT (Medium Chain Triglycerides)** on food labels.

People with MCADD will also benefit from a **diet that has fewer of the total calories provided in the form of (any) fat than usual**, since much of it cannot be used, and fasting should be avoided. Their diet should have the same calories any baby needs ... and the baby with MCADD will communicate that quite well in most cases. **What it does mean is that a lower proportion of calories would come from fat and a higher proportion would come from carbohydrate than is typical in the usual American baby diet.**

[The fish oil supplementation described earlier is not calculated in the “grams of fat goal per day,” primarily because it is a very small amount, and it is a form of very long-chain fat that people with MCADD could burn better than other fats if they sent it to the furnace to be burned for energy. However, the biggest reason to include some is because there are a number of important construction projects that require these fats (including brain structure) and I want to assure that they have them on hand. In other words, they are not being given just for their caloric value as a fuel source.]

Breastfeeding babies with MCADD should be encouraged for many reasons.

Interestingly, mother’s milk has a generous amount of fat in the form of MCT because it is easy for babies to digest. This does not appear to be primarily related to the forms of fat the mother eats ... she just seems to make them from other fats and ships them off to make milk.

One small adjustment may be helpful: It is reasonable for the mother who is nursing her baby with MCADD to avoid using coconut oil regularly while she is nursing. It is not known if this source of MCT would alter the amount in a mother’s milk ... since she seems to make her own MCT for the milk anyway ... but it seems reasonable and generally not a huge problem to simply avoid using coconut oil until baby is weaned.

Special formulas for babies with MCADD are available.

If a baby is going to be fed formula, there are special mixtures that can provide all the calories and nutrients of regular formula. They can be made to have less total fat and, in particular, no medium-chain fat. Some mothers choose to mostly breast-feed with a feeding or two from a special fat-free / higher carbohydrate formula. **In this situation, a pediatric nutritionist needs to be involved to assure that nothing baby needs is left out, and that nothing baby should not have is accidentally left in.**

Include lots of brightly colored fruits and vegetables in the diet of anyone with MCADD ... and also in the diet of anyone without MCADD. ☺

Anyone with a metabolic disease (like MCAD or diabetes or PKU or many more) has a **higher need for antioxidants** to protect against a higher than usual production of potentially injurious “free radicals.” We all produce free radicals all the time, but altered metabolism clearly results in a much higher production of them. This higher free radical production can contribute to a wide variety of health issues, so higher free radical production makes one need a higher than usual intake of antioxidant substances to protect against injury to cells.

Happily, a generous intake of brightly colored fruits and vegetables will provide terrific and potent antioxidants, so this goal is not difficult to accomplish. They are also almost always very low in fat, so they are “freebies” for a person with MCADD. Eating more of these foods benefits everyone, but helping a person with unusual metabolism to regularly eat these foods is particularly beneficial. Additionally, the person with MCADD may also have a beneficial effect on the health of the rest of the family since all will be eating more fruits and veggies!



(“Eat the Rainbow!”)

Miscellaneous things to keep an eye on:

Monitor the baby’s weight, length and head circumference progress (which should all be in the normal range ... there is nothing about MCADD that should result in a growth pattern different from that expected for others in a family.)

Evaluate if the baby seems to be much hungrier than expected for little people of his/her age, especially if the hunger is unusual considering the amount of food eaten.

In that situation, the baby may also often be seen to be putting on a lot of **extra body fat** but still acting hungry. It may be that the **form** of calories needs to be adjusted to provide more as carbohydrate and provide less as fat that can’t be burned and ends up being stored. I call this the “Baby Fluffiness Index.” ☺

Bottom Line:

Wherever you live, be sure that you find a team of health care professionals that can serve as a resource to helping to manage your baby’s diet. MCADD is rare but there are people in every state who specialize in unusual metabolic problems. If your baby has MCADD, they will help with all of this every step of the way and be sure that you learn all about it so your baby will be safe and thrive. If you happen to come to see us here at Sanford Medical Center in Fargo, ND, please do call or email me if any questions or concerns about your baby’s special diet arise. All my contact information is in the upper right corner. Don’t be shy ...