



## Newborn Screening FACT Sheet

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# Elevated C3-DC Acylcarnitine Malonic Acidemia (MAL)

### What is MAL?

Malonic Acidemia (MAL) is a condition in which the body is unable to break down certain proteins. It is classified as an organic acid condition because MAL can lead to harmful levels of organic acids and toxins in the body. Signs and symptoms of the condition vary. However, early identification and treatment can improve the health of children with MAL.

### What Causes MAL?

When we eat food, enzymes help break it down. Some enzymes help break down fats into their building blocks, called fatty acids. Other enzymes break down these fatty acids for energy. In MAL the enzyme malonyl-CoA decarboxylase is not working correctly. This enzyme's job is to help break down fatty acids for energy. If your baby has MAL, his or her body does not make enough or makes non-working malonyl-CoA decarboxylase enzymes. When this enzyme does not work correctly, your baby's body cannot break down fatty acids for energy. If fatty acids cannot be broken down, it causes heart trouble (the heart needs fatty acids for energy) and a build-up of harmful substances in the body.

### What Symptoms or Problems Occur with MAL?

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

There are very few reported cases of MAL. According to these reported cases, signs can start any time from just a few days after birth to 13 years of age. Usually, if your baby does not receive preventative treatment, signs of MAL will start in early childhood. These signs include:

- developmental delays
- weak muscle tone (called hypotonia)
- diarrhea
- vomiting
- seizures

Many of these signs may occur when your baby eats foods that his or her body cannot break down. They can also be triggered by long periods of time without eating, illnesses, and infections.

If your baby shows any of these signs, be sure to contact your baby's doctor immediately.

### What is the Treatment for MAL?

**Dietary Treatments** - Your baby may need a restricted diet to avoid high-fat foods. A dietician or nutritionist can help you plan the right diet to keep your baby healthy while still giving him or her nutrients needed for growth and development.

Eating often will also help prevent your baby from experiencing many of the signs mentioned in the "Symptoms or Problems" section. Illnesses and infections can also trigger these signs.

**Supplements and Medications** - Your doctor might prescribe L-carnitine supplements to help break down fats and to remove harmful substances from the body. L-carnitine is a natural substance, but your baby's body might not make enough of it. You will need to get a prescription from your baby's doctor for these supplements.

### Things to Remember

Because MAL is so rare, the outcomes are not well known. MAL can cause an enlarged, weak heart (also called cardiomyopathy), so you may need to work with a cardiologist to monitor your child's heart. The dietary and supplement treatments mentioned in the Treatment section are likely to help children live much longer, but there has been no research conducted on children with MAL who receive treatments.

Without treatment, children could die during infancy.