TEXAS Department of State Health Services

Newborn Screening FACT Sheet

Medium-Chain Ketoacyl-CoA Thiolase Deficiency
(MCAT or MCKAT)

What is MCAT?
Medium-chain ketoacyl-CoA thiolase deficiency (MCAT) is a condition in which the body is unable to breakdown certain fats. It is considered a fatty acid oxidation condition because people affected by MCAT are unable to change some of the fats they eat into energy the body needs to function. This can cause too many unused fatty acids to build up in the body. MCAT can cause vomiting, liver problems, and death if left untreated. The effectiveness of treatment is unknown.

What Causes MCAT?
When we eat food, enzymes help break it down. Some enzymes break down fats into their building blocks, called fatty acids. Other enzymes break down these fatty acids. The enzyme medium-chain ketoacyl-CoA thiolase helps break down certain fatty acids. Fatty acids are built like chains and come in a variety of lengths. They are categorized as either short, medium, long, or very long. Medium-chain ketoacyl-CoA thiolase helps break down medium length fatty acid chains.

In MCAT deficiency, this enzyme is not working correctly. It may be that the body does not make enough of the enzyme or that the body makes non-working enzymes. When this enzyme does not work, the medium length fatty acids cannot be broken down for energy. Fatty acids are an important source of energy for the heart, especially when sugars are low, such as in between meals. When fatty acids are not completely broken down, harmful substances build up in the body. MCAT causes dangerously high levels. While everyone has some acid in his or her blood, high levels can be toxic.

We do not yet know the genetic basis of MCAT.

What Symptoms or Problems Occur with MCAT?
[Symptoms are something out of the ordinary that a parent notices.]

In a reported case of MCAT, the baby began showing signs two days after birth. The signs of MCAT included:

- vomiting
- weight loss
- poor appetite
- diarrhea
- trouble breathing

What is the Treatment for MCAT?
In a case of MCAT, the baby was treated with IV fluids to lower the acid levels in his blood. While everyone has some acid in his or her blood, high levels can be toxic.

Things to Remember
Medium-chain ketoacyl-CoA thiolase deficiency is extremely rare. A baby with this condition died at 13 days of age.