

Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency General Overview

Q. What is MCAD deficiency?

A. MCAD deficiency is a treatable disorder that affects the way the body breaks down fats. If left untreated, MCAD deficiency can cause life-threatening illness.

Q. How does the body normally process fats?

A. The body normally uses carbohydrates and sugars from our diet for energy and uses fats as an energy reserve. When all of the carbohydrates and sugars in our bodies have been used, we break down fats for energy. One of the enzymes that helps break down fats is called MCAD.

Q. What happens to fats in a child with MCAD deficiency?

A. In a child with MCAD deficiency, fats cannot be broken down normally because the MCAD enzyme does not work properly. This can result in high levels of partially broken down fats, which are toxic to the brain and nervous system.

Q. What are the effects of having MCAD deficiency if it is not treated?

A. Untreated MCAD deficiency can result in mental retardation, other problems of the nervous system and sometimes death.

Q. What is the treatment for MCAD deficiency?

A. Treatment for MCAD deficiency is usually straightforward. Treatment involves avoiding long periods of time without eating and having meals that are high in carbohydrate and low in fats (breast milk and commercial formula are fine). Infants should have at least one night-time feeding, or a late night snack, to reduce the time they go without eating. Special care must be taken if a person with MCAD deficiency becomes ill and has trouble keeping food down. This is usually treated in the hospital with an intravenous feeding. It is important that children with MCAD deficiency receive specialized management through a clinic with experience in treating this disorder.

Q. Why would a child have MCAD deficiency?

A. MCAD deficiency is an inherited disorder. It results when a baby receives a double-dose of a non-working MCAD gene (one from each parent). For more information about this, contact your health care provider or a genetic counselor.

Q. What are the chances that a child will be born with MCAD deficiency?

A. About one in every 20,000 babies in the United States is born with MCAD deficiency. However, parents who have a child with MCAD deficiency each carry one copy of the non-working gene. That means, with each pregnancy, there is a one in four chance of the child having MCAD deficiency.

For more information about MCAD deficiency, please see the Disorders section or our website: www.doh.wa.gov/nbs.

