Carnitine uptake defect (CUD)

**CAUSE**

CUD occurs when an enzyme, called “carnitine transporter” (CT), is either missing or not working properly. This enzyme’s job is to carry a substance called carnitine into our cells. Carnitine helps the body make energy from the fat in food and also helps us use fat already stored in the body. When the normal CT enzyme is not functioning properly, the body must rely solely on glucose. There is a limited supply of glucose in our bodies and once it is used up, the body tries to use fat without success. This leads to low blood sugar, called hypoglycemia, and the buildup of harmful substances in the blood. This is referred to as a metabolic crisis.

**IF NOT TREATED**

There are two main forms of CUD: one begins in infancy and the other in childhood. Babies with CUD usually begin showing symptoms between birth and three years old. Metabolic crisis often occurs when a child has not eaten for more than a few hours, gets sick, or develops an infection. Repeated episodes of metabolic crisis can cause brain damage and result in learning problems or mental delays.

**TREATMENT OPTIONS**

Your child will need to be under the care of a metabolic specialist and dietician. Treatment is usually needed throughout life.

- Your child needs to avoid going a long time without food. This is to avoid a metabolic crisis. These children should not go more than 4 to 6 hours without food and some may require more frequent feedings. It is important that these children be fed in the night meaning you will need to wake them up to eat if they do not wake up on their own and even if they are not hungry.
- The dietician will develop a diet for your child that contains low amounts of fat and a high amount of carbohydrates. Your child’s diet should never be changed without the approval of your dietician.
- The main treatment for CUD is a supplement of L-carnitine. This is a safe and natural substance that helps body cells make energy. It also helps the body get rid of harmful wastes. L-carnitine can reverse the heart problems and muscle weakness that occur in children with CUD.
- Your metabolic specialist must prescribe the L-carnitine supplement and instruct you on the recommended dose.
- Contact your child’s doctor immediately at the start of any illness. Children with CUD need to be treated in a hospital to prevent serious health problems.

**IF TREATED**

With prompt and careful treatment, children with CUD usually live healthy lives with typical growth and development. Infants with CUD who have repeated episodes of metabolic crisis may have permanent brain damage. This can cause learning disabilities or mental delays.