

Carnitine Supplementation and VLCAD Deficiency

There has been considerable controversy regarding the role of L-carnitine ([Carnitor®, Sigma-Tau Pharmaceuticals, Inc.](#)) and its value as a supplemental therapy in fat oxidation disorders. Currently, there is concern that it could actually be harmful with VLCAD Deficiency. There is actually no concrete evidence for this concern; in fact, there has been much experience that indicates that there is no harmful aspect of L-carnitine in patients with VLCAD Deficiency. It is now known that VLCAD Deficiency can present in three very different ways. These phenotypes should be recognized when discussing the question of carnitine supplementation in this inherited disorder.

1. Some children with VLCAD present identically to MCAD Deficiency-Hypoglycemia (VLCAD-H).
2. Some present as recurrent rhabdomyolysis (muscle breakdown like adult onset CPTII Deficiency). These patients are usually young adults or teenagers (VLCAD-M).
3. Many children present as the cardiomyopathic form and have often died in the first year of life (VLCAD-C). They also have problems with blood sugar and muscle is also involved.

VLCAD-H seems to only involve liver metabolism. VLCAD-M involves skeletal muscle only, whereas VLCAD-C involves heart, muscle and liver.

All have been treated with L-carnitine supplement and increased concentrations of medium-chain triglyceride (MCT) containing formulas or diets. In our experience, there has not been any untoward effect of L-carnitine, including heart rhythm disturbances, in any of these patients even during acute illnesses during which intravenous carnitine was administered to patients with the cardiomyopathic form of the disease.

The reason we use L-carnitine in VLCAD Deficiency is to maintain a reasonably normal ratio of Free Co-enzyme A to esterified Coenzyme A to facilitate mitochondrial metabolism during fat

mobilization due to fasting or unexpected illness.

For further discussion of the direct experience with long-chain fat oxidation disorders, including VLCAD, please feel free to contact us. For those who have had a different direct experience with L-carnitine and a patient, we would be interested in the details.

Dr. Charles R. Roe

Dr. Alfred Slonim