

Description	Natural History	Genotype-Phenotype Correlation	Treatment	Prognosis
Classic MCAD deficiency	<p>If undiagnosed, up to 25% mortality with first metabolic crisis</p> <p>Hepatomegaly, accompanied by hypoketotic hypoglycemia, hyperuricemia, elevated LFTs, mild hyperammonemia, elevated anion gap</p> <p>May have developmental regression, aphasia, ADHD, chronic muscle weakness</p>	<p>Intrafamilial phenotypic differences suggest environmental factors may also play a role in the natural history of the disorder</p> <p>Most commonly homozygous for the K304E (*%A>G) mutation</p>	<p>Avoidance of fasting</p> <p>Emergency letter</p> <p>Prompt interventions with IV glucose</p> <p>Carnitine supplementation in certain cases</p>	<p>Frequent feedings of infants</p> <p>Avoidance of fasting</p> <p>Consider uncooked cornstarch at bedtime for toddlers</p> <p>Relatively low fat diet (>30% of energy from fat)</p> <p>AVOIDANCE of medium chain triglycerides</p>
"Mild" MCAD deficiency	<p>Milder abnormalities on acylcarnitine profile</p>	<p>Individuals should be considered at risk for developing clinical symptoms</p> <p>Either compound heterozygotes for K304E and another mutation or homozygous for other mutations (Albers et al, 2001; Zschocke et al, 2001)</p>	<p>Avoidance of fasting</p> <p>Emergency letter</p> <p>Prompt interventions with IV glucose</p> <p>Carnitine supplementation in certain cases*</p>	<p>Frequent feedings for infants</p> <p>Avoidance of fasting</p> <p>Consider uncooked cornstarch at bedtime for toddlers</p> <p>Relatively low fat diet (>30% of energy from fat)</p> <p>AVOIDANCE of medium chain triglycerides</p>

- Carnitine supplementation is controversial. Treem et al (1989) found no improvement in fasting tolerance with carnitine supplementation. Lee et al (2005) have recently demonstrated improved exercise tolerance with short term supplementation.

Clinical Classification of MCAD Deficiency