MEDIUM-CHAIN Acyl CoA DEHYDROGENASE DEFICIENCY (MCAD)

MCAD deficiency is the most common fatty acid oxidation disorder. MCAD generally presents clinically between the second month and second year of life, although presentation may occur from 2 days to 6 years. MCAD results in recurrent episodes of hypoglycemia, acidosis, seizures, vomiting and coma when an affected infant is stressed. Early detection, prevention of fasting and monitoring for illness or infection are essential to prevent death in infants with MCAD.

Prevalence:	1:10,000- 1: 15,000 live births
Analyte Measured:	Acylcarnitines
Reporting Ranges:	Elevated octanylcarnitine (C8-Cn)
Feeding Effect:	None
Timing Effect:	Specimens collected within the first 24 hours of life may be elevated.
Other Effect:	None
Confirmation:	Frequent feedings are important in preventing symptoms in an infant who may have MCAD. Repeat filter paper specimen, blood and urinary metabolites and confirmation by DNA analysis for A985G mutation, as directed by program staff or Metabolic Specialist. **Imposed fasting is not recommended as a screening test or confirmation of the disorder.**
Treatment:	Referral will be made to a Metabolic Specialist. Avoidance of fasting and monitoring of illness or infection. Supplementation with L-carnitine may be indicated. Prompt treatment of illness and possible intravenous glucose therapy may be indicated when unable to maintain oral intake.

Comment:

Affected children are healthy and usually asymptomatic until stressed by fasting, illness or infection. Clinical symptoms include lethargy, vomiting, encephalopathy, respiratory arrest, hepatomegaly, seizures, apnea and cardiac arrest.

Symptoms which are triggered by fasting or infection are fatal in 25% of undiagnosed infants with first event. MCAD may be misdiagnosed as Reye Syndrome and may be responsible for 2-7% of SIDS deaths.

Maine Newborn Screening Program
207-287-5357 or 1-800-698-3624