

**FACT SHEET**  
Healthcare Provider

**Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)**

**Description:**

Medium Chain Acyl-CoA Dehydrogenase Deficiency is an autosomal recessive genetic condition. MCADD is caused by a defect in the MCAD enzyme, one of the enzymes involved in the mitochondrial beta-oxidation cycle. Individuals with this disorder are unable to break down fatty acids for use as an alternative energy source. Metabolic crisis can occur during periods of fasting and can cause episodes of hypoglycemia, vomiting, coma, and even death.

**General Population Incidence:**

1:25,000 live births

**Symptoms:**

Metabolic crisis may be precipitated by intercurrent illnesses and may present very quickly in infants/children who are not feeding properly. MCADD typically presents with hypoketotic hypoglycemia, lethargy, vomiting, seizures, coma or sudden death. Infants may be more subject to sudden death than older children. About 20% of infants with MCADD die during an acute episode when the diagnosis is not yet suspected. Survivors may suffer from neurological sequelae such as developmental delay, seizures, attention deficit hyperactive disorder or other behavioral abnormalities.

**Diagnosis:**

Newborn screening--Tandem mass spectrometry identifies elevations in medium chain acylcarnitines (C8-C8/C10 ratio).

The Newborn Screening Laboratory may request a second dried-blood-spot filter paper card if the initial screening result is above the normal range. Infants with presumptive positive screening (critical) results require prompt follow up. If this occurred, the clinician would be contacted by the Metabolic Treatment Center. When notified of these results, the clinician should immediately check on the clinical status of the baby and facilitate referral to the Metabolic Treatment Center. The Metabolic Treatment Center will provide consultation and assistance with diagnostic testing.

**Situations that risk metabolic decompensation:**

Children with MCADD are often clinically asymptomatic. Metabolic decompensation can be triggered by the catabolic processes that occur in the course of an infection, after an immunization or with a prolonged period of fasting.

**Monitoring:**

Clinical observation is the most important tool for monitoring patients with MCADD. They should be observed for recurrent vomiting, refusal to eat, increased lethargy, apnea or seizures. In these situations, immediate evaluation in the emergency room is necessary. In situations of metabolic decompensation hypoglycemia can develop, but a normal blood glucose does not rule out metabolic instability and should never be a reason to delay therapy. It is also important for the primary care provider and the Metabolic Treatment Center to develop an on-going collaborative relationship in caring for these patients.

**Treatment:**

- Avoid fasting
- Feed at regular intervals during the day and limit overnight fasting.
- Should not go without food intake longer than 4 hours for the first 4 months of life; 6 hours for ages 4-8 months; and no longer than 8 hours thereafter.
- Restriction of dietary fat is controversial but it is reasonable during intercurrent infections; The Metabolic Treatment Center will set a patient's diet prescription that determines the optimum percentage of fat, carbohydrate, and protein.
- Carnitine supplements are provided in the case of a low blood carnitine level.
- Increased carbohydrate intake and cornstarch therapy is necessary during acute illness. The use of cornstarch therapy is an ongoing treatment and is even more necessary during acute illness.
- If the child is vomiting or refuses to eat, the child needs to be taken to an emergency room for IV administration of at least 10% dextrose.
- Infants and children with MCADD should have regularly scheduled visits at the Metabolic Treatment Center.
- The parents should have an emergency protocol with them at all times. This protocol can be provided by the Metabolic Treatment Center, and contains basic information about the disorder, necessary diagnostic investigations and guidelines for treatment.

**Illness:**

- Any illness can potentially lead to metabolic decompensation.
- Prevention and /or early intervention are of particular importance.
- Consult with the Metabolic Treatment Center within 24 hours of the onset of the illness or at the time of hospitalization.
- Provide high-carbohydrate feedings including cornstarch.
- Avoid dehydration.
- Closely monitor blood glucose and intake; even if blood glucose is normal, metabolic decompensation can occur.

**Immunization:**

- Immunizations must be kept current.
- All children with MCADD should have a yearly vaccine for influenza.
- There is no contraindication to any immunization because of MCADD.
- Parents and physicians should be alerted to the need for immediate evaluation if high fever, lethargy or vomiting occurs in the first 24 hours.
- After an immunization without any other clinical symptoms, administration of acetaminophen or ibuprofen is warranted.

**Surgical/surgical procedures:**

- Discuss any plans for surgical and dental procedures with the Metabolic Treatment Center
- Infants and children with MCADD can undergo necessary anesthetic/surgical procedures.
- Any surgical procedure constitutes a potentially catabolic situation.
- Any surgery should include hospitalization pre- and postoperatively.
- Preoperative fasting should be avoided, with 10% dextrose being started preoperatively and continuing postoperatively until the child is eating and drinking well.
- If complicated surgery, or a postoperative period, as an inpatient is required, the procedure should be done at a hospital with a metabolic service.

**Growth and development:**

- It is crucial to closely monitor all growth parameters on a regular basis.
- In cases with neurological deficits, the child should be referred to an early intervention program and developmental progress closely monitored by both the metabolic team and the primary care provider.



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November 2005