



State of CT Genetics Newborn Screening Program Health Care Provider Fact Sheet

Carnitine Palmitoyltransferase Deficiency (CPT I)

Carnitine Palmitoyltransferase Deficiency (CPT I) is a very rare metabolic disorder that affects the skeletal muscles and their ability to function properly. CPT Deficiency belongs to a group of diseases that involve the defective metabolism of lipids. Prolonged periods of strenuous exercise can trigger an episode of symptoms. These may include muscle aches, stiffness, and weakness. Muscle tissue may break down (rhabdomyolysis) and abnormal levels of myoglobin may be present in the urine (myoglobinuria). The urine may appear dark reddish-brown after prolonged periods of exercise. Disabling muscle weakness, stiffness, and pain may last for days. CPT Deficiency is more apparent in individuals with diabetes and those who may be malnourished.

Clinical Features

Initial symptoms have occurred with or after episodes of fasting, infection or diarrhea and usually presents between 8-18 months, but a few have presented in the first week of life. Hypoketotic hypoglycemia, hyperammonemia, lethargy, vomiting and seizures proceed to coma and death if not recognized and treated aggressively. Hepatic dysfunction with elevated ammonia and enzymes is common. Creatine kinase elevation may be seen in acute episodes, although myoglobinuria is usually absent. No evidence of chronic muscle weakness has been documented. Renal tubular acidosis was seen in several patients. Transient hypertriglyceridemia was also seen.

The classical findings of hypoketotic hypoglycemia without dicarboxylic aciduria and with a high plasma carnitine level—both total and free—distinguish CPT I deficiency from the other known defects in the pathway.

Recurrent episodes are common and generally have been successfully treated with glucose infusion and have been avoided by preventing fasting. Frequent feeding and replacement of dietary long chain fat with MCT have been beneficial.

Diagnosis

Newborn screening—Tandem mass spectrometry—Free carnitine (C0)—elevated/normal, C0/C16 ratio very elevated, C0/C18 ratio very elevated

Confirmation—a second sample may be requested or follow up testing will be done at the Metabolic Treatment Center at Yale or UCONN Genetics.

Situations that risk metabolic decompensation

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. Lethargy, vomiting, apnea or seizures are typical clinical features with or without hypoglycemia. Infants may be more subject to sudden death than older children.

Monitoring

Clinical observation is the most important tool for monitoring these patients. 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
- The Metabolic Treatment Center will set a patient's diet prescription that determines the optimum percentage of fat, carbohydrate, and protein.
- Supplementation with Medium Chain Triglycerides Oil (MCT)
- 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, (s)he needs to be taken to an emergency room for IV administration of 10% dextrose.
- The parents usually have an emergency protocol with them. This protocol, provided by the Metabolic Treatment Center, contains basic information about the disorder, necessary diagnostic investigations and guidelines for treatment.
- These infants and children should have regularly scheduled visits at the Metabolic Treatment Center.

Illness

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

Immunization

- Immunizations must be kept current.
- All children with this disorder should have a yearly vaccine for influenza.
- There is no contraindication to any immunization.
- 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 this disorder 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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860-509-8081

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