



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

3-Methylcrotonyl-CoA Carboxylase Deficiency (3MMC or 3MCC)

Introduction

3-Methylcrotonyl-CoA Carboxylase (3-MCC) deficiency is an autosomal recessive disorder of leucine catabolism. It produces an accumulation of 3-Methylcrotonyl-CoA. The clinical course in the untreated infant has been quite variable. The onset of symptoms is generally after 3 months of age, but can be variable. Many individuals have no symptoms into adulthood. Some infants have presented with a Reye-like illness with hypoketotic hypoglycemia, metabolic acidosis and liver dysfunction often precipitated by an intercurrent illness, which has led to fulminant liver failure and death in some cases. Others present with muscle hypotonia and failure-to-thrive in conjunction with recurrent episodes of vomiting and diarrhea. In general, the earlier the presentation the poorer the prognosis.

Diagnosis

Newborn screening—Tandem mass spectrometry: C5OH

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 infections, after an immunization, increased physical activity or with a prolonged period of fasting.

Monitoring

Clinical observation is the most important tool for monitoring patients with 3MCC. They should be observed and assessed for neurological status, 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

- Low-protein diet with restricted leucine intake
- The Metabolic Treatment Center will set a patient's diet prescription that determines the optimum percentage of fat, carbohydrate, and protein.
- The parents should have an emergency protocol with them at all times. This protocol, provided by the Metabolic Treatment Center, contains basic information about the disorder, necessary diagnostic investigations and guidelines for treatment.
- Infants and children with 3MCC 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
- Care should be coordinated by the Metabolic Treatment Center

Immunization

- Immunizations must be kept current, but patients and physicians should be alerted to the need for immediate evaluation if high fever, lethargy, or vomiting occurs in the first 24 hours.

Surgical/surgical procedures

- Discuss any plans for surgical and dental procedures with the Metabolic Treatment Center.
- A surgical procedure constitutes a potentially catabolic situation and preoperative fasting should be avoided with 10% dextrose being started preoperatively and continuing postoperatively until the child is eating and drinking well. Any procedure requiring anesthesia 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.
- Intellectual prognosis depends on early diagnosis and treatment and, subsequently, on compliance with the dietary and supplement plan.



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