

## FATTY ACID OXIDATION DISORDERS

### CARNITINE UPTAKE DISORDERS (CUD)

Carnitine, a natural substance in food is used by cells to process fats to produce energy. Infants with primary Carnitine deficiencies have proteins called carnitine transporters that do not work so the body can not use fat for energy particularly during periods without food (fasting). Initial signs and symptoms are lethargy, enlarged and poor pumping heart, vomiting, muscle weakness, and low blood sugar. If left untreated infant is at risk for heart failure, coma, and sudden death.

#### Screening Method:

Use of Tandem Mass Spectrometry to measure carnitine level.

#### Treatment:

Carnitine supplementation and avoidance of fasting relieves episodes of hypoglycemia and is curative.

#### Incidence:

1:200,000 births

### LONG -CHAIN HYDROXYACYL-CoA DEHYDROGENASE (LCHAD) DEFICIENCY

Deficiency of LCHAD impairs conversion of dietary and fats stored in the body to energy. Signs and symptoms in the newborn are failure to thrive and cardiac problems. Death may occur from heart and respiratory or liver failure if identification and treatment are delayed.

#### Screening Method:

Identification of elevated levels of hydroxy acylcarnitines (C16-OH, C18:1-OH) by Tandem Mass Spectrometry method.

#### Incidence:

1:50,000 births

### MEDIUM CHAIN ACYL- CoA DEHYDROGENASE (MCAD) DEFICIENCY

This disorder is caused by the lack of MCAD enzyme that breaks down fats stored in the body into sugars used to produce energy for bodily functions. Infants are usually asymptomatic but can go into shock or die suddenly if the infant has problems with feeding for more than 4 hours.

#### Screening Method:

Use of Tandem Mass Spectrometry to measure levels of Octanoylcarnitine (C8 acylcarnitine) and Carnitine esters.

#### Treatment:

Avoid fasting for more than 4 hours as infant. Glucose supplementation and hydration required during illness. Food grade cornstarch mixed in liquid should be taken

at bedtime to help decrease morning low blood sugar.

Incidence:

1: 10,000 births

#### TRIFUNCTIONAL PROTEIN DEFICIENCY (TFP)

Mitochondrial TFP is a condition that prevents the body from converting certain fats to energy, particularly during prolonged fasting and/or periods of increased energy demands such as fever or stress. Signs and symptoms of this disorder may begin in infancy or later in life.

Symptoms that occur during infancy include feeding difficulties, lack of energy, low blood sugar, weak muscle tone, and liver problems. Infants are at risk for heart problems, breathing difficulties, coma, and sudden death.

Screening Method:

Tandem Mass Spectrometry is used to detect elevations of several long chain and hydroxy acylcarnitines (C18:1-OH), C16-OH, C14-OH, C18).

Treatment:

Supportive care for acutely ill child involves IV fluids containing glucose and bicarbonate. Administration of L-Carnitine should be considered. Avoid fasting and certain types of food. It is recommended that parents travel with a letter of treatment guidelines from pediatric metabolic disease specialist.

Incidence:

Not Known

#### VERY LONG -CHAIN ACYL-CoA DEHYDROGENASE (VLCAD) DEFICIENCY

The deficiency of the VLCAD is one of 4 mitochondrial enzymes that begins the process of converting fatty acids to energy particularly during periods without food. Signs and symptoms typically appear during infancy or early childhood and include low blood sugar, lack of energy, muscle weakness, and are at risk for serious complications such as liver abnormalities and heart problems. This disorder is sometime mistaken for Reye syndrome.

Screening Method:

Use of Tandem Mass Spectrometry to detect increased levels of long chain acylcarnitines (C14:1, C16,C18).

Treatment:

Treatment includes a high carbohydrate low fat diet, special formula, regular feeding, and avoiding strenuous exercise.

Incidence:  
1:200,000 births