UHealth UNIVERSITY OF MIAMI HEALTH SYSTEM | MILLER SCHOOL OF MEDICINE UNIVERSITY OF MIAMI

EMERGENCY REGIMEN V 1.0 Bo 1-24-12

Patient label

Primary Geneticist: _____

On call UM Geneticist Tel: 305-331-3023

Protocol seen and approved: _____

Medium Chain Acyl-CoA Dehydrogenase Deficiency

The above named patient has Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD). MCADD is an inborn error of fatty acid metabolism where medium chain fatty acids are not broken down properly. As a consequence fatty acids cannot be readily converted to glucose and/or ketone bodies when needed during episodes of increased energy demand or when glycogen stores are exhausted. Toxic metabolites may accumulate. Blood glucose levels may remain normal despite a severe metabolic decompensation. Infants with MCADD are at risk for hypoglycemia, liver dysfunction (Reye-like syndrome) and sudden death during episodes of decompensation. Routine treatment for MCADD involves a high-carbohydrate, low fat diet and the avoidance of prolonged fasting. L-carnitine may be supplemented.

Affected patients with MCADD are therefore at greatest risk for metabolic decompensation when one or more of the following are present:

1) **Intercurrent illness in particular when food/fluid intake is less than 75% of normal**
2) **Recurrent vomiting and/or diarrhoea**
3) **Prolonged fasting**
4) **Weight loss of more than 10%**
5) **High fever**
6) **Trauma and/or surgery**

The following symptoms may be a sign of metabolic decompensation:

Drowsiness, lethargy, refusal to feed, vomiting, seizures, profuse sweating, changes in behavior

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MINOR ACUTE ILLNESS (No fever, upper airway infection, otitis)

1) Increase fluid intake using sugar drinks (Gatorade, Apple Juice..) or tea with maltodextrin (20g/100ml)

2) Supplementation with L-carnitine to 50 mg/kg/day may be considered. Please consult metabolic physician before doing so.

3) Re-assess situation after 8 hours; if situation worsens, have patient evaluated in pediatric ER

ACUTE ILLNESS (Fever, vomiting…) EMERGENCY PROTOCOL

1) Assess the patient clinically
   If the patient is unconscious or lethargic immediately start infusion with 10% glucose (see under 3) followed by blood sampling. Do not delay the infusion!

2) Secure an intravenous line and check glucose immediately (AccuCheck)

3) When hypoglycemia (<45 mg/dl) is present or patient shows severe symptoms inspite of being normoglycemic, give 25% glucose 2ml/kg iv push, followed by a continuous 10% glucose/0.33% NaCl supplemented with KCl as indicated at 1.5X maintenance. Target blood glucose level are 80-130 mg/dl. Hyperglycemia should be avoided due to the risk of lactic acidosis. Correct with insulin if needed.

4) Draw bloods for CBC, blood gas, e'lytes, lactate, ammonium, liver enzymes, CK, free and total carnitine, acylcarnitine profile, organic acids in urine (UM Metabolic laboratory) and ketones in urine, additional tests as indicated

5) Treat any underlying illness or symptom (infection, dehydration, vomiting) as indicated

6) A supplement with L-carnitine 100mg/kg/day divided in 3 doses is recommended by some experts. Please consult metabolic physician before L-carnitine supplementation.

7) **Avoid the following medications: Salicylates, fat binding/producing anesthetics (f.ex. propofol), steroids. Intralipids are contraindicated!**

8) **Immediately notify the on-call UM Geneticist at 305-331-3023. This is a 24/7 service**

Disclaimer: the above recommendations cannot replace an individual medical evaluation by a board certified physician. The UM Geneticist on-call should always be informed. UM is not responsible in case the protocol has not been followed.

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