

Illness Protocol Example For Long-Chain Hydroxy-Acyl-CoA Dehydrogenase (LCHAD) Deficiency

Clinical Presentation:

Rhabdomyolysis, Anorexia, Vomiting, Lethargy, +/- Hypoglycemia, +/- Cardiomyopathy, +/- Retinopathy

Routine Treatment:

Diet: Portagen for infants or MCT oil supplements for older children and adults. High Carbohydrate, restricted fat 25-30% of total calories. Avoid Fasting longer than 6-8 hours ~ eating often throughout the day is important for any child/adult with an FOD

Medication: Carnitor® (100 mg/ml) solution give 50 mg/kg/day, divided in 3-4 doses

Minor Acute Illness without Fever (Ear infections, URI, etc...):

Diet: As above plus, increase fluid intake - Gatorade, Powerade, Pedialyte, Popsicles, Jello, Apple juice

Medication: Carnitor® (100 mg/ml) solution give 50 mg/kg/day, divided in 3-4 doses

Acute Febrile Illness associated with significant anorexia, +/- vomiting, diarrhea:

Bloodwork: CBC with differential
Complete Metabolic profile
CPK
Dry bloodspot card for Acylcarnitine profile
Urine OA

Treatment:

Medications: Carnitor® (100 mg/ml) solution give 100 mg/kg/day, divided in 3-4 doses. If not tolerated switch to IV Carnitor®

Loading dose: IV Carnitor® (1000 mg/5 ml), 50 mg/kg slowly over 2-5 minutes, followed by 50 mg/kg over each subsequent 24 hour period, until the patient is able to resume their normal PO diet

IV D10 1/4 NS @ 1 ¼ - 1 ½ maintenance to maintain blood sugar ~100 mg/dl

**** Intralipids are CONTRAINDICATED in patients with Fatty Acid Oxidation Disorders**

Acute Rhabdomyolysis:

If CPK \geq 25,000 and rising, recommend ICU admission for combined IV Glucose and Insulin drip to stop further lipolysis. Monitor blood CPK and Glucose levels.

***** www.fodsupport.org has more information on LCHAD and other FODs**