

EMERGENCY REGIMEN

Patient label

Primary Geneticist: _____

On call UM Geneticist Tel: 305-331-3023

Protocol seen and approved: _____

Isovaleric acidemia

The above named patient has **Isovaleric acidemia**.

Isovaleric acidemia is an inborn error of metabolism where the amino acid leucine is not broken down properly. In the presence of catabolism due to infection or substantially reduced food intake, patients may have ketosis, metabolic acidosis and neurological symptoms. Metabolic management may include a leucine-restricted diet, carnitine supplementation and intensified emergency treatment during acute episodes of intercurrent illness.

If this patient presents with illness, please immediately notify the on-call UM Geneticist at 305-331-3023. This is a 24/7 service.

Affected patients with Isovaleric acidemia are 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 diarrhea
- 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

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) Continue supplementation with L-carnitine at an increased dose 100 mg/kg/day orally.
- 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) 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
- 3) Treat any underlying illness or symptom (infection, dehydration, vomiting) as indicated
- 4) L-carnitine should be provided PO (100-200 mg/kg/day divided TID) or IV (200 mg/kg/day).
- 5) Treat metabolic acidosis aggressively with IV Sodium bicarbonate (1mEq/kg). Treating conservatively in the expectation of a re-equilibration of acid/base balance as other biochemical/clinical parameters are normalized can lead to tragic consequences.
- 6) **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.