EMERGENCY METABOLIC DISEASE PROTOCOL

This individual is part of a family with a history of late onset ornithine transcarbamylase deficiency, an inherited metabolic disorder of the urea cycle characterized by the accumulation of ammonia and glutamine in the blood and in the brain. If affected, males in this family can experience lethargy progressing to coma and death. The disorder worsens in case of fasting or fever, when the body needs to use endogenous proteins. **Do NOT give steroids as they cause catabolism and can cause an acute decompensation.**

In cases of **acute decompensation**, we recommend the following:

1. **The Emergency Room physician should see the individual immediately.**
2. Stop protein intake.
3. Obtain the following laboratory studies: basic metabolic panel, plasma ammonia, plasma amino acid, liver function tests.
4. Give the following **IV fluids** to provide calories and decrease ammonia:
   - D10 Normal Saline 20 mEq/L KCl at 1.5 times maintenance.
   - Goal sodium is greater than 150. If necessary, give 3% saline at 1 mL/kg x 1 dose. Maintaining NA > 150 will help reduce brain edema associated with hyperammonemia.
   - IV Mannitol (1g/kg per dose) can be given if there are signs of cerebral edema.
5. Intralipids 20% at 1/10th the rate of the IV fluids
6. Arginine 1 g IV every 8h (up to 150-200mg/kg/day)
7. If ammonia is above 150, consider admission to the intensive care unit. Ammonia should be repeated every 4 h until normalized.
8. Glucose levels should be checked every 2 h, with a target glucose level between 120 and 170 mg/dL. Insulin may be started (0.1 U/kg bolus followed by the same dose per h as a drip) if glucose is >200 mg/dL.
9. If vomiting or prior to scavenger therapy (below), consider using Ondasentron (0.15 mg/kg, up to every 8 hours PRN).
10. It may be necessary to begin treatment with IV ammonul (benzoate/phenylacetate) at 55mL/square meter if unconscious or with ammonia >200. Keep this medication separate from the maintenance fluids. If using Ammonul, use D10 (without saline) for maintenance fluids.

Please contact us immediately by calling 801-662-1000 and ask for the Medical Geneticist on call to be paged.

Thank you in advance,

[Signature]

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