A Brief Discussion on the Identification and Care of Type I & III GSD Patient in the Emergency Room

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Glycogen Storage Diseases

- This group of diseases is characterized by the accumulation of normal or abnormal glycogen due to deficiency of the enzymes of glycogen metabolism. Although rare, they have contributed greatly to the understanding of glycogen metabolism.
- It presents in both adults and children, in infants commonly 3-4 months.

Clinical and Laboratory Findings

- Type I: May develop hypoglycemia (lack of glycogenolysis by epinephrine or glucagon), hyperlipidemia, hyperuricemia and lacto acidosis.
  - Often hypoglycemia is missed due to presence lactic acid.
- Also known as von Gierke's disease
- In infants, they present at 3-4 months of age with hepatomegaly.

Case Study

- GSD is rarely, if ever diagnosed in the ER.
- 24 year old male presents to the ER complaining of:
- Dizziness
- Sweating
- Nervousness
- Palpitations

Identification and Diagnosis

Initial reaction of ER Staff should be to:

- Stop
- Look
- Listen
- Feel
- Labs

Identification

- Stop: Slow down
- Look: For Medical Alert Bracelets, Cards in Wallet, I.C.E. contact information in mobile. Evidence of Corn Starch ingestion in and around the lips and mouth.
- Listen: If accompanied by family member, friend etc, listen carefully to what information they convey and also to patient, if they have the ability to articulate.
  - Be prepared that patient or his/her advocate will be far more knowledgeable on the subject than yourself.
- Feel: Palpitate liver (use of ultrasound)
- Labs: Blood sugar, triglycerides, Cholesterol, pH, P_{CO2}, BBG, electrolytes and lactates.

Treatment

Upon suspicion of GSD in the adult, treatment should begin immediately, including:

- IV fluids with D10½, normal saline at 125cc's (12.5 gm/hr glucose). Glucose concentrations should be checked hourly to ensure hypoglycemia is not occurring.
- D10 should not be weaned until patient is able to tolerate his usual regimen of dietary intake and cornstarch.

The Pediatric Patient

- Children tend to present with acidosis.
- Type I patients generally present with lactic acidosis and hypoglycemia
- In Type III, VI & IX patients present with ketonacidosis
  - On assessment you can find weakness, lethargy, tremors, nausea and vomiting, convulsions, coma
  - Blood gas findings of decreased pH, decreased bicarbonate, decreased base excess, decreased P_{CO2} (if lung compensation)

Treatment of the Pediatric Patient

- For Children less than 3 years of age:
  - IV fluids with D10 ¼ , normal saline at 1.25 to 1.5 X maintenance.
  - Glucose concentrations should be checked hourly to ensure hypoglycemia is not occurring.
  - Give enough glucose to reverse acidosis

- For children over 3 years of age:
  - IV fluids with D10 ¼ to D10 ¼ to ½ , normal saline at 1.25 to 1.5 maintenance.
  - Glucose concentrations should be checked hourly to ensure hypoglycemia is not occurring.

- In pediatric patient, dietary management, including frequent high-carbohydrate meals with cornstarch supplements or nocturnal gastric drip feedings are usually effective.

- D10 is to be weaned.

Warning

- Lactated Ringer's should not be given.

- Glucagon should never be administered to a patient with GSD as it will lead to severe acidosis.

- Patient must be weaned.

Discharge

- GSD is rarely, if ever diagnosed in the ER.
- On discharge, the patient should be referred to the appropriate specialist for follow up.
- Specific instructions on diet, until specialist evaluates on follow up.

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Overview

- Carbohydrate metabolism a vital role in cellular function by providing the energy required for most metabolic processes.
- Glucose is the principal substrate of energy metabolism in humans. Metabolism of glucose generates ATP via glycolysis in mitrochondrial oxidative phosphorylation.
- Dietary sources of glucose are obtained by polysaccharides, primarily starch and disaccharides including lactose, maltose and sucrose.
- Galactose and Fructose are 2 other monosaccharides that provide fuel for cellular metabolism; however, their role as fuel sources is much less significant than that of glucose.

Overview continued

- Galactic is derived from lactose (galactose + fructose) which is found in milk products, and is an important component for certain glycolipids, glycoproteins and glycosaminoglycans.