



Management of Hypoglycemia in Von Gierke's Disease - A Case Study

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Introduction

- Glycogen storage disease type-1 (GSD-1), or Von Gierke disease, is a rare phenomenon primarily affecting the liver and kidney
- Deficiency in glucose 6-phosphatase (G6P) or microsomal transport proteins, results in excessive accumulation of glycogen, along with fat in the liver, kidney, and intestinal mucosa
- Patients with GSD-1 have a wide spectrum of symptoms including hepatomegaly, hypoglycemia, lactic acidemia, hyperlipidemia, hyperuricemia, and growth retardation
- Manifestations of the disease vary in age of onset, rate of disease progression, and severity [1]

Case Presentation

- An 18-year-old male with GSD-1, presented to the hospital with hyperuricemia, hypertension, nausea, and vomiting
- Patient followed an hourly cornstarch regimen during the day and overnight through infusion via PEG tube
- The complaints started at work where he was unable to tolerate oral cornstarch
- He was hemodynamically stable on arrival. ABG showed pH 7.372, PaCO₂ 30.3, PaO₂ 92.2. WBC 16.80, K⁺ 5.8, HCO₃ 13, BUN 28, Cr 2.2, glucose 60, AST 115, ALT 128, cholesterol 352, triglycerides >1000, uric acid 10.6, and lactic acid 11.8, which trended down to 8.0.

- CT abdomen showed hepatomegaly and fatty infiltration with PEG tube in place
- He was admitted to the ICU and was started on D5NS for hypoglycemia and lactic acidosis. Per request by the patient's pediatrician, he was transitioned to IV D10/0.45NS at 110mL/Hr to maintain a blood glucose above 75 mg/L.
- Frequent accuchecks were done till he could tolerate his dietary regimen with cornstarch
- Lactic acid down trended to 2.9 and accuchecks ranged between 100-110. Cr improved to 1.3 and he was discharged in stable condition with plans for a further genetic therapy work up.

Discussion

- Mainstay therapy for Von Gierke disease is prevention of metabolic derangements for which dietary and lifestyle changes are recommended
- A low fructose and sucrose diet is recommended by limiting the intake of galactose and lactose to one serving per day [2][3]
- Hypoglycemia treatment in such patients is two fold, by utilizing both quick and stable release sources
- Cornstarch has been once such therapy since the 1980s; its slow digestion provides a steady release of glucose over a longer period of time, as compared with other sources of carbohydrates.
- Dosing guidelines vary from age and person, but it is highly recommended to check BG levels frequently to maintain a BG > 70 mg/dL. Associated high levels of triglycerides and cholesterol can be treated with statins, fibrates, etc.

Conclusion

- The management of hypoglycemia in GSD-1 disease presents various obstacles which could prove to be fatal
- Due to the deficiency of G6P, treatment with a specialized hypoglycemic regimen is warranted
- A D10 ½ NS infusion can be used to maintain blood sugar levels as well as correct metabolic or lactate imbalances
- Infusion should be gradually weaned off after the patient can tolerate oral feeds, as this can help prevent the risk of hypoglycemia and other derangements
- Further research is needed in regards to these patients for more sustainable regimens

References

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