

A Rare Case of Hyperlactatemia in The Emergency Department

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Abstract

Glycogen storage disease type 1a is a rare autosomal recessive syndrome characterized by hypoglycemia, hyperuricemia, hyperlipidemia, hepatomegaly, among other features. Case report: A 31-year-old woman genetically diagnosed with this disease in childhood was admitted to the Emergency Department with tachypnea. Her arterial lactate was 179 mg/dL, bicarbonate of 2 mmol/L, pH of 7.0 and pCO₂ 2.2 mmHg. She received IV glucose, isotonic bicarbonate, and antibiotics. Her urine culture was positive for *Escherichia coli*. She had a complete recovery from acidosis in 12 hours and was discharged three days later. Conclusion: This case highlights a rare differential of lactic acidosis that can, sometimes, be present in the Emergency Department.

Key Words: glycogen storage disease, von Gierke disease, hypoglycemia, lactic acidosis, emergency medicine.

Introduction

Hyperlactatemia occurs when lactate production exceeds lactate consumption. Lactic acidosis is an anion gap metabolic acidosis that can be classified into two types: those with tissue hypoxemia (type A) and those without tissue hypoxemia (type B)^{1,2}.

Type B etiologies include intoxication (i.e. metformin), inborn errors of metabolism (i.e. glycogen storage disease), diabetic ketoacidosis, malignancy, and alcoholism¹.

Glycogen storage disease (or glycogenosis) refers to a group of hereditary metabolic disorders with improper metabolism of glycogen, resulting in hypoglycemia and lactic acidosis. Depending on the type of disease, autosomal inheritance can be dominant or recessive³.

We report a case of glycogen storage disease type 1a presenting with type B hyperlactatemia in the Emergency Department (ED) and performed a literature review about this rare condition.

Case Report

A 31-year-old woman presented to the ED with a two-day history of tachypnea. She denied fever, cough, and other systemic symptoms.

In her first year of life, she was genetically diagnosed with glycogenosis type 1a (also known as von Gierke disease). She was using atorvastatin, ciprofibrate, and a special corn starch diet for her condition.

At admission, she was afebrile, heart rate was 120, blood pressure of 120/76 mmHg, oxygen saturation was 99%, and respiratory rate of 36 ipm with good peripheral perfusion.

Workup exams demonstrated an arterial pH of 7, bicarbonate of 2 mmol/L, base excess -28 mmol/L, lactate 179 mg/dL, anion gap of 30, pO₂ 118 mmHg, pCO₂ 2.2 mmHg, mild elevation of inflammatory markers, and leukocytosis of 15.820/mm³. She promptly received IV glucose (250 mg/kg followed by 3-4 mg/kg/min), isotonic 8.4% bicarbonate (1 mmol/kg/h), and ceftriaxone (2g daily) to prevent hypoglycemia, manage acidosis and treat a potential infection, respectively.

Exams to determine the cause of decompensation demonstrated a urine culture positive for *Escherichia coli*. Blood cultures and a thorax computed tomography were negatives. As a result, we continued to use ceftriaxone for cystitis treatment.

We decided to start an oral diet after twelve hours (Table 1) when the acidosis was resolved. She was discharged three days later.

The patient provided informed written consent for publication.

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Received: 17.11.2021 • **Accepted:** 13.01.2022

DOI: 10.33706/jemcr.1003145

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Cite this article as: Stumpf MAM, Simões AL, Alencar JCG. A rare case of hyperlactatemia in the emergency department. Journal of Emergency Medicine Case Reports. 2022;13(1): 29-30

Table 1: Evolution in exams parameters during hospital stay

	Bicarbonate (mmol/L)	Lactate(mg/dL)	Glucose (mg/dL)
Admission*	2	179	87
2 hour	5.2	-	140
4 hours	6.9	-	-
8 hours*	8.6	259	220
10 hours	14.3	257	-
12 hours	21	257	180
20 hours	25.5	-	-
40 hours	20	23	-

*From arterial blood, the others samples are from venous blood

Discussion

As previously described, glycogenosis is a rare genetic disorder characterized by an inborn glycogen metabolism error. Type 1a is defined by a deficit in glucose-6-phosphatase (G6Pase) enzyme, which catalyzes the final phase of glycogenolysis and gluconeogenesis with glucose-6-phosphate transporter (G6PT). Type 1b, however, is caused by a G6PT mutation. The disease is autosomal recessive, with an overall incidence of 1:100 000 cases, with about 80% of these having type 1a³.

The symptoms differ depending on the patient's age. In most cases, the diagnosis is made during childhood. In the neonatal period, doll-like facies, hypoglycemia, lactic acidosis, and hepatomegaly may be present. Hypoglycemia, lactic acidosis, hyperlipidemia, hypertriglyceridemia, and hyperuricemia are among the conditions that can occur during evolution. Type 1b has neutropenia and reduced neutrophil functions, but type 1a has a functional immune system. Inflammatory bowel disease is also more common in type 1b^{3,4}.

The diagnosis is based on clinical features and confirmation of mutations in the G6Pase and SLC37A4 (G6PT) genes⁵. Liver biopsy is not mandatory; however, a diagnosis may be made by measuring the G6Pase enzyme activity in a piece of snap-frozen liver biopsy tissue³.

Management is based on appropriate food intake to prevent hypoglycemia – such as corn starch. Fructose and galactose metabolism is impaired when the G6Pase enzyme is deficient, hence these sugars should be avoided or minimized. Multivitamins and calcium may be required because of the restricted diet^{3,6}.

Allopurinol may be used for hyperuricemia as well as statins and fibrates for hyperlipidemia. For type 1b neutropenia, a granulocyte colony-stimulating factor is considered first-line therapy. For patients who develop microalbuminuria, ACE inhibitors should be initiated along with oral bicarbonate or citrate to treat persistent acidosis^{3,6}.

There is limited evidence in the literature for acute management. We followed the recommendation in the last European Guideline: a bolus of 250 mg glucose/kg followed by 3-4 mg/kg/min and IV bicarbonate for acidosis correction⁶. There is no report about the optimal concentration for bicarbonate repositioning or when the oral diet should be reintroduced. We opted for isotonic bicarbonate concerning the sodium level and restarted the oral diet once the acidosis was under control and the patient's tachypnea had improved. In cases of acute decompensation, it is important to search for precipitating factors including dietary non-compliance and bacterial or virus infections².

The issue with acute decompensation is respiratory fatigue rather than acidosis. To our best knowledge, this is the second case report of glycogenosis presented in the ED. The first case was described by Oster et al. in 2016. In their report, the patient had a catastrophic evolution caused by respiratory failure and required orotracheal intubation. Even dialysis was considered for acidosis management since sodium level was too elevated as a result of bicarbonate administration².

Conclusion

Glycogenosis is a rare cause of lactic acidosis, mainly in the pediatric population. The management should include IV glucose for hypoglycemia prevention and IV bicarbonate for acidosis control.

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