The multiple manifestations of Thiamine deficiency

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Introduction

Thiamine (B1) deficiency most commonly manifests as Beriberi disease or Wernicke's encephalopathy. It can also present with gastrointestinal symptoms; however, this is usually underrecognized. Rarely, it can produce severe illness. Herein, we present a case of patient that presented with multiple manifestations of thiamine deficiency.

Case Presentation

A 46-year-old female presented to our hospital complaining of abdominal pain. The pain had been present for 9 months, however intensified 10 days prior. It was localized in the epigastrium, was sharp, non-radiating, and exacerbated by food intake. The pain was associated with nausea and non-bloody vomiting. Additionally, she had numbness on both upper and lower extremities for the same period of time. She stated drinking alcoholic beverages on a daily basis, usually wine and gin. Her diet consisted of toasts. She was not taking any medications or herbal supplements.

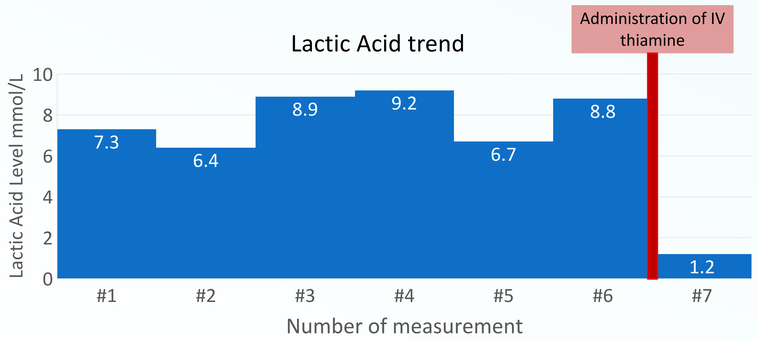
Upon presentation, her heart rate was 117 beats/min, blood pressure was 145/ mmHg, temperature 37.1 °C, and oxygen saturation of 100% breathing room air. On physical exam, she had epigastric tenderness. She also had decreased light touch and pin prick sensation on her extremities, worst on the distal region and extended proximally to her elbows. She had no motor deficits or abnormal eye movements on physical exam.

Diagnosis and Treatment

Blood work was significant for Hb of 11 mg/dl and MCV 104 femtoliters/cell. Lactic acid (LA) was 7.3mmol/L at presentation, and LA trend is presented in Table 1. Due to the elevated LA septic workup was done, however no source of infection was identified on blood cultures, urine analysis or chest Xray. A computed tomography angiography of the abdomen was done which ruled out bowel ischemia. Prior to current admission, an upper endoscopy had been normal. Lastly, a B1 level was checked and was significantly low (<6nmol/L) confirming the diagnosis.

After receiving IV fluids, her symptoms remained unchanged. Subsequent treatment with IV infusion of thiamine 250 mg daily resulted in complete normalization of LA (Table 1). Her gastrointestinal symptoms also improved after two days of IV B1 therapy. Her numbness persisted and Gabapentin was added with mild improvement of her symptoms.

Table 1



Discussion

B1 is a required cofactor for pyruvate dehydrogenase and α-ketoglutarate dehydrogenase (a-KDGH) ; key enzymes for glucose metabolism. In B1 deficiency, a-KDGH activity is reduced, which ultimately shunts the metabolism of glucose to lactate. It also results in decreased production of myelin that can lead no peripheral neuropathy.

B1 deficiency can result from inadequate diet, impaired liver storage, excessive requirements, and inborn enzymatic defects. Risk factors also include alcohol abuse, gastric bypass, and systemic diseases.

Deficiency usually manifests as Wernicke encephalopathy or Beriberi disease. An under-recognized gastrointestinal syndrome of B1 deficiency has been described. This syndrome consists of nausea, vomiting, abdominal pain and lactic acidosis, was first described in 2004, and is known as gastrointestinal beriberi.

Conclusion

B1 deficiency leads to decreased activity of a-KDGH, which in turns shift glucose metabolism to lactic acid; it also results in decreased production of myelin. Here we present a patient that had three different manifestations of thiamine deficiency: Unexplained lactic acidosis, gastrointestinal symptoms, and peripheral neuropathy (dry Beriberi). Most of her symptoms improved after IV thiamine treatment. Delayed diagnosis of B1 deficiency as a cause of elevated lactate and gastroenterological symptoms can lead to unnecessary tests and procedures like this patient. Physicians should have high suspicion in patients with history of alcohol intake.

1. Manzanares W, Hardy G. Thiamine supplementation in the critically ill. *Curr Opin Clin Nutr Metab Care.* 2011;14(6):610–7. doi: 10.1097/MCO.0b013e32834b8911.
2. Donnino M. Gastrointestinal beriberi: a previously unrecognized syndrome. *Ann Intern Med.* 2004;141(11):898–9. doi: 10.7326/0003-4819-141-11-200412070-00035.
3. Manzetti S, Zhang J, Van der Spoel D. Thiamin function, metabolism, uptake, and transport. *Biochemistry.* 2014;53(5):821–35. doi: 10.1021/bi401618y.
4. Butterworth RF. Thiamine deficiency-related brain dysfunction in chronic liver failure. *Metab Brain Dis.* 2009;24(1):189–96. doi: 10.1007/s11011-008-9129-y.
5. Donnino MW, Vega J, Miller J, Walsh M. Myths and misconceptions of Wernicke's encephalopathy: what every emergency physician should know. *Ann Emerg Med.* 2007;50(6):715–21. doi: 10.1016/j.annemergmed.2007.02.007.
6. Andersen LW, Mackenhauer J, Roberts JC, Berg KM, Cocchi MN, Donnino MW. Etiology and therapeutic approach to elevated lactate levels. *Mayo Clin Proc.* 2013;88(10):1127–40. doi: 10.1016/j.mayocp.2013.06.012.