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Case Report

Von Gierke Disease (Glycogen Storage Disease Type I) and Life-Threatening Abdominal Aortic Aneurysm: A Case Report of an Extremely Rare Condition

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Von Gierke disease, also known as glycogen storage disease type I, co-existent with an abdominal aortic aneurysm (AAA), is an extremely rare combination of diseases that requires challenging therapeutic measures. We present, for the first time in literature, the case of a 62-year-old female with von Gierke disease who required open surgical repair of an AAA with challenging neck anatomy outside of instructions for use of endovascular repair. Even though the surgical risks for life-threatening complications, such as pancreatitis, metabolic acidosis, and kidney failure, were high, the 6-month postoperative course was uneventful. Despite the invasiveness of the treatment, surgery to treat the AAA was safe and effective. Further data is needed to draw robust conclusions about the treatment of choice for those patients with diseases in co-existence with AAAs.

Key Words: Glycogen storage disease, von Gierke disease, Abdominal aortic aneurysm, Open repair

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INTRODUCTION

Glycogen storage disease type I (GSDI), better known as von Gierke disease, is a rare hereditary disease that affects one in 100,000 of the population. It consists of a deficiency of enzymes that play a significant role in glycogen metabolism [1].

This syndrome manifests with hypoglycemic episodes, lactic acidemia, hyperlipidemia, and neutropenia (GSDIb). Additionally, in the long term, these patients may present with kidney failure and hepatic malignancies [2]. When these patients require major surgery necessitating general anesthesia, they are at high risk of perioperative hypoglycemia, metabolic acidosis, rhabdomyolysis, myoglobinuria, acute renal failure, pancreatitis, and excessive bleeding [2,3].

The co-existence of this disease with an abdominal aortic aneurysm (AAA) is an extremely rare occasion that poses a serious therapeutic challenge, considering the perioperative and postoperative difficulties associated with an aortic repair. This case report aims to highlight the demanding peri- and postoperative management with increased rates of complications, and the absolute necessity of a multidisciplinary approach in order to achieve the best medical care for these patients.

This case report adheres to the ethical principles outlined in the Helsinki Declaration, including the principles of informed consent, confidentiality, and protection of patient privacy. The patient provided written informed consent for

anonymized information, including images, to be published in this article. Since the manuscript was a case report based on an observational study of a patient, Institutional Review Board approval was not required.

CASE

A 62-year-old female, previously diagnosed with GSDI, was presented to our outpatient care with the sonographic finding of an asymptomatic AAA measuring 5.6 cm in diameter. Hypertension, hepatomegaly, kidney dysfunction, and hyperuricemia were included as comorbidities. As an indication for surgical treatment was evident, a computed tomography angiography (CTA) was performed (Fig. 1). By evaluating the CTA images, we concluded that the characteristics (reverse tapered anatomy, infrarenal angulation >60°, diameter of the infrarenal neck <22 mm) were outside of the instruction for use for an endovascular aneurysm repair (EVAR); thus, from the morphological point of view, the open repair approach was preferred [4]. A fenestrated endovascular aneurysm repair (FEVAR) and chimney endovascular aneurysm repair (CHEVAR) approach was also considered, but neither option was selected since these complex endovascular techniques require a greater amount of contrast agent and thus increase the risk of acute renal failure.

On the other hand, the diagnosis of von Gierke disease made the required anesthesia for open surgical repair quite challenging due to the described risks [2,3]. Preoperatively, the patient underwent fibrate therapy for three days in order to achieve lower triglyceride levels and minimize the risk of postoperative pancreatitis. Additionally, dextrose solution was administered the day prior to the operation, and the aneurysm was successfully excluded by the placement of an aortoiliac Y-prothesis. The operation lasted two hours, with an overall aorta clamping time of 40 minutes. During the operation, minimal blood loss (400 mL) was observed. Additionally, a minimal dosage of catecholamines was administered. No hypoglycemic crisis or acidosis was noted perioperatively. The patient was successfully extubated, had palpable peripheral pulses, and stayed in the intensive care unit (ICU) for two days. During this time, the monitoring of glucose, lactate, and base excess through regular arterial blood analysis revealed normal-range glucose levels; this eliminated the possibility of a hypoglycemic or acidosis episode. A constant glucose perfusion was performed until the glucose levels were stable within the range of 90-120 mg/dL. Following that, Nutriflex, a nutritional solution consisting of lipids, carbohydrates, and amino acids totaling 765 kcal per liter, was administered through an IV at 40 mL/h. Lipofundin, containing medium-chain triglycerides and glycerol with a total caloric intake of 1,980 kcal/L, was also administered at 11 mL/h. Since the patient was 150 cm tall and weighed 80 kg, the body mass index was calculated at 35.6 kg/m^2; therefore, a total of 750 kcal for the first 12 hours was determined to be sufficient nutrition.

After two days in the ICU, the patient returned to the peripheral vascular station for four additional days. During the entire in-hospital postoperative period, the patient required transfusion of three units of erythrocyte concentrates; meanwhile, the triglyceride values decreased from 1,600 mg/dL to 1,052 mg/dL and therefore plasmapheresis

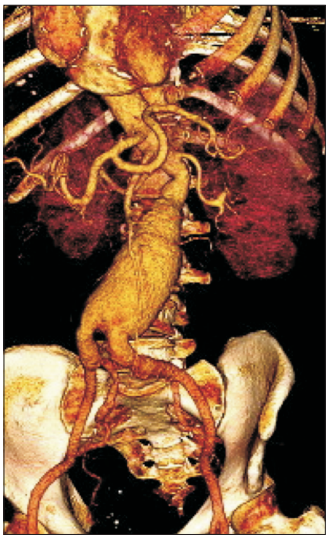

Fig. 1. Preoperative computed tomography angiography showed the reverse tapered anatomy, infrarenal angulation and small diameter of infrarenal neck.

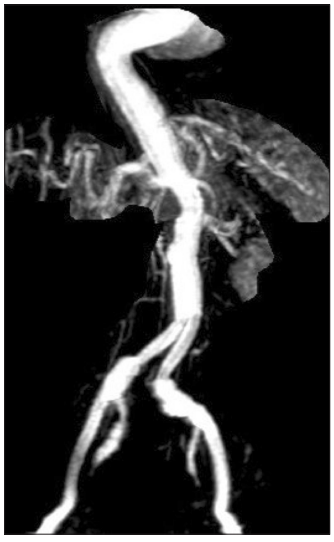

Fig. 2. Postoperative magnetic resonance tomography angiography showed the exclusion of the aneurysm and stenosis-free flow to the groins.

was not necessary. Additionally, a temporary increase of 0.5 mg/dL in the creatinine levels was observed, reaching a maximum of 1.6 mg/dL; this was treated with adequate hydration, resulting in the reversion of creatinine levels to the normal range. On the day of discharge, the glomerular filtration rate (GFR) was 89 mL/min, the Hb level was >9 g/dL and the patient was fully mobilized, free of symptoms, and in an overall good condition.

The 6-month follow-up was uneventful. A magnetic resonance tomography angiography of the abdomen showed exclusion of the aneurysm and stenosis-free flow to the groins (Fig. 2).

DISCUSSION

GSDI is a rare disorder with an incidence of one in 100,000 [1]. Meanwhile, AAA in females is an uncommon disease, with an estimated prevalence of 0.43% in females at the sixth decade of life [5].

To our knowledge, this is the first case in literature with the co-existence of GSDI and a life-threatening AAA, necessitating a novel approach to treatment. Our literature review revealed a limited number of articles [2,3,6-20], while the majority of articles described liver or kidney transplants in children or young adults. We could only identify three cases with patients in the range of 40-60 years of age and none above 60 years [6-8].

Two of the most prominent articles were case series with 80 and 6 patients, respectively, where the patients suffered from GSDIa or GSDIb and required major surgeries [2,3]. Gurrieri et al. [3] in a series of six patients reported two intraoperative hypoglycemias that were treated with dextrose solutions and three perioperative metabolic acidosis that resolved after 24 hours. No postoperative pancreatitis or any other adverse event was reported. On the other hand, Boers et al. [2] in their study of 80 patients, reported four postoperative deaths (one suicide, one malignancy, one transplant rejection, and one death due to acute pancreatitis and sepsis two months after the surgery). They also reported eight acute and six chronic kidney failures; however, only three patients required dialysis. After reviewing the literature, it becomes apparent that surgery is associated with intraoperative hypoglycemic episodes and metabolic acidosis, as well as postoperative renal failure. Since this is the first documented case with combined GSDI and AAA, the vascular status of these patients has not yet been documented. As their triglyceride levels remain extremely high for their entire lives, it is quite logical to expect increased vascular tissue frailty with damaged arterial walls and an increased risk of thromboembolization. However, during the operation, the arterial wall of the abdominal aorta was similar to that of any other patient suffering from AAA, disproving this hypothesis.

Although our first choice of treatment was EVAR, the reverse tapering and the narrow and angulated infrarenal neck excluded the possibility of a minimally invasive treatment with conventional EVAR. Additionally, taking into consideration the risk of potential renal failure for a patient with GSDI, the nephrotoxicity of the contrast agent used in the more complicated endovascular techniques (FEVAR and CHEVAR) was another deterring factor of the endovascular approach.

On the other hand, the open approach hides the risk of intraoperative hypoglycemic episodes as well as acute postoperative pancreatitis. Even though we administered fibrates to counter hyperlipidemia, triglyceride values were constantly >1,200 mg/dL, further increasing the risk of acute pancreatitis. After a multidisciplinary meeting between the vascular and anesthesiology departments of our hospital, accompanied by a specialist for metabolic disorders, we decided to proceed with an open repair. A temporary creatinine spike, which reached 1.6 mg/dL and was resolved with hydration after 48 hours, was the only observed event. During the stay at the hospital, the metabolic status was stable and within normal ranges.

Based on our experience with postoperative management, we suggest regular monitoring of blood glucose levels and the provision of more frequent meals in order to minimize the risk of hypoglycemic episodes. In cases where feeding is not feasible, we suggest using appropriate parenteral nutrition. We also suggest regular monitoring of creatinine levels and adequate hydration. The use of fibrates or other cholesterol-lowering medications cannot be suggested since they did not appear to affect triglyceride values.

In conclusion, GSDI and life-threatening AAA is an extremely rare combination of diseases that impose a serious therapeutic challenge. The comorbidities and intraoperative and postoperative risks that come with GSDI are a prominent factor in therapeutic decision-making but should not be given more importance than any other factor. The anatomical characteristics of the aneurysm should dictate the surgical technique and the decision to perform an open or EVAR.

A multidisciplinary approach is required in order to establish a peri-, intra-, and postoperative plan with all the above-mentioned measures so that the risks of an adverse event can be minimized.

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CONFLICTS OF INTEREST

The authors have nothing to disclose.

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Concept and design: AGP, KPD. Analysis and interpretation: AGP. Data collection: FD, MK. Writing the article: AGP. Critical revision of the article: AGP, KPD, NAB. Final approval of the article: all authors. Statistical analysis: none. Obtained funding: none. Overall responsibility: AGP.

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