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(CASE REPORT)

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Pre-operative anesthesia management of a child patient with Cystinosis

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Abstract

Cystinosis is an autosomal recessive lysosomal storage disease caused by CTNS gene mutation, characterized by abnormal accumulation of cysteine amino acid. Its prevalence is approximately between 1:100,000 and 1:200,000. It may affect internal organs such as the cornea, bone marrow, thyroid, liver and kidneys. It is divided into three groups: infantile nephropathic, juvenile nephropathic and ocular cystinosis.

There are limited reports in the literature regarding the peroperative management of these patients. We wanted to focus on the peroperative anesthesia management of our patient who was diagnosed with cystinosis and was planned to undergo emergency testicular torsion surgery.

Keywords: Cystinosis; Anesthesia management; Perioperative care; Lysosomal Storage Diseases

1. Introduction

Cystinosis was first described in 1903 by the Emil Abderhalden as the familial cystine accumulation disease [1]. It is an autosomal recessive lysosomal storage disease caused by CTNS gene mutation, characterized by abnormal accumulation of cysteine amino acid. Its prevalence is approximately between 1:100,000 and 1:200,000 [2].

The cystinotic process may affect various tissues and internal organs such as the cornea, conjuctiva, bone marrow, leukocytes, brain, muscle, thyroid, spleen, liver and kidneys. The kidneys are initially affected during the first year of life through proximal tubular damage followed by progressive glomerular damage [4].

Three clinical patterns have been described in patients with cystinosis including the infantile nephropathic form, the juvenile nephropathic form, and the ocular nonnephropathic form.The most frequent and most severe form of cystinosis is infantile nephropathic cystinosis [5].

In this case, we wanted to focus on the peroperative anesthesia management of our patient, who was diagnosed with infantile nephropathic cystinosis when he was 8 months old.

2. Case report

Our patient is a 13 year old male, weighing 35 kg. The patient was referred to pediatrics by his family at the age of 8 months with complaints of growth retardation, polyuria and constipation. He was diagnosed with Fanconi syndrome and infantile nephrropatic cystinosis after tests. He is currently receiving Cystagon (Cysteamine) 150 mg, Kalinor (potassium bicarbonate, citrate) and Phosphorus treatments.

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The patient's preoperative laboratory findings were normal except for Potassium is 3.1 mEq/L. The patient was consulted to pediatrics. A detailed explanation was given to the the patient's relatives and consent was taken. The patient was given ASA III-E and emergency testicular torsion surgery was planned. Considering respiratory system and renal complications, he was taken into surgery with postoperative intensive care monitoring.

The patient was taken to the operating room after standard ECG, noninvasive blood pressure, temperature and saturation monitoring. Anesthesia was induced with 0.02 mg/kg midazolam, 1-2 mg/kg profopol and 1 mcg/kg fentanyl. A size 2.5 LMA was placed. Anesthesia was maintained with 1.5 MAC Sevoflurane in 50-50% oxygen air. The operation was completed after 70 minutes, and the LMA was replaced from the patient under deep sedation. Postoperative analgesia was provided by acetaminophen. After the patient was monitored in the postoperative unit, he was taken to the intensive care unit. The patient, whose respiratory and metabolic status was stable, was transferred to the urology service after 1 day of intensive care follow-up. After 2 days of urology service follow-up, the patient recovered and was discharged.

3. Discussion

Cystinosis was first described in 1903 by the Emil Abderhalden as the familial cystine accumulation disease [1]. It is a rare autosomal recessive lysosomal storage disease characterized by abnormal accumulation of cysteine amino acid. Its prevalence is approximately between 1:100,000 and 1:200,000 [2]. The responsible gene is CTNS gene which , is located on the short arm of chromosome 17, encodes the carrier protein cystinosin that transporting cystine out of the lysosomal compartment [3]. The frequency of occurrence of the mutation type may vary depending on geographical location.

The cystinotic process may affect various tissues and internal organs such as the cornea, conjuctiva, bone marrow, leukocytes, brain, muscle, thyroid, spleen, liver and kidneys. The kidneys are initially affected during the first year of life through proximal tubular damage followed by progressive glomerular damage. The latter results in Fanconi's syndrome and renal insufficiency, often necessitating the need for kidney transplant. Renal losses of calcium and phosphate may lead to bony abnormalities and fracture also [4].

Three clinical patterns have been described in patients with cystinosis including the infantile nephropathic form, the juvenile nephropathic form, and the ocular nonnephropathic form.The most frequent and most severe form of cystinosis is infantile nephropatic cystinosis. Children with infantile nephropatic form appear normal at birth, but demonstrate failure to thrive by around 6–9 months of age. Symptoms of kidney dysfunction develop such as polyuria and polydipsia by 6–18 months of age [5]. The nephropathic juvenile form is diagnosed in the minority of the patients and most of the patients are diagnosed older than 10 years, and the ocular nonnephropathic form manifests only with photophobia and blepharospasm due to cystine accumulation in the cornea [6].

The supportive and symptomatic treatment of cystinosis aims to maintain fluid, electrolyte and acid-base balance, provide nutritional support, prevent the development of rickets and ensure adequate levels of necessary hormones. [2]. Beyond supportive treatment, the only specific therapy for cystinosis is cysteamine (β-mercapto-ethylamine). Cysteamine depletes lysosomal cystine by cleaving free cysteine and cysteamine–cysteine mixed disulfide, which is exported from lysosomes. Cysteamine was first used in cystinosis patients in 1976 but wasn't approved by the FDA until 1994 [7,8]. Although effective in delaying complications of cystinosis, cysteamine therapy cannot treat established renal Fanconi syndrome. Oral cysteamine treatment cannot prevent the accumulation of corneal cysteine, but topical cysteamine applied 10-12 times a day is effective after months.

As cystinosis has multiorgan involvement, it has several anesthetic implications, although the issue of primary concern is underlying renal involvement. Important to the perioperative care is the avoidance of potentially nephrotoxic agents and attention to intravascular volume and renal blood flow. Many drugs used in anesthesia routines are excreted by renal. Although two metabolic products of sevoflurane, fluoride ions and compound A, were thought to be nephrotoxic when administered at high concentrations for long periods with low fresh gas flow, subsequent studies have shown that sevoflurane preserves renal function even in patients with renal failure [9,10]. The induction agents (propofol, fentanyl, and lidocaine) undergo hepatic metabolism and exhibit limited renal clearance.

Approximately 24% of cystinotic patients may experience myopathy. The myopathy often begins distally with weakness and wasting of hand muscles and progresses to more generalized involvement. Myopathies may become life-threatening as involvement of the respiratory muscles [11]. Patients with clinical myopathy may require postoperative mechanical ventilatory support. Neuromuscular monitoring should be used in cases where relaxant is being used [12].

Considering the normal renal function tests and nephrotoxicity, we preferred induction with propofol midazolam and fentanyl, maintenance with sevoflurane, and analgesia with acetaminophen. We didn't use the neuromuscular blockers.

Cystinosis has not previously been reported to mimic malign hyperthermia. The patient could place at risk of iatrogenic hyperthermia when routine intraoperative precautions against hypothermia are instituted. Progressive orthopaedic involvement and growth failure may also affect perioperative care. Poor nutrition and bony abnormalities may make these patients prone to develop pressure sores. In addition, the general cachectic nature of these patients may place them at risk for intraoperative hypothermia. Considering all these situations, patients should be monitored with a temperature probe [13].

4. Conclusion

Cystinosis is among the rare diseases. Anesthetic management of a patient with cystinosis should focus on determining the extent of end organ involvement during preoperative evaluation. The primary concern during perioperative care is to preserve renal function and maintain fluid-electrolyte balance. Nephrotoxic drugs should be avoided and all drugs should be given in titrated doses.

Progressive orthopedic involvement and growth failure may affect perioperative care. Malnutrition and bone anomalies can facilitate the development of pressure sores. Careful filling of bone protrusions is recommended. The cachectic nature of patients may cause the risk of intraoperative hypothermia. Warming infused fluids, regulating operating room temperature, and humidifying respiratory gases will help prevent significant drops in body temperature. Taking all these situations into consideration, patients should be monitored with a temperature probe.

Various anesthesia-related complications have been reported in the literature. Preoperative evaluation should include all systems to ensure appropriate perioperative care can be provided.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed.

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