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Case report

Anaesthetic management of a child with cystinosis

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ABSTRACT

Cystinosis is a lysosomal storage disorder which is characterized by abnormal accumulation of amino acid cysteine. Cystinosis affects various tissues of the body and has several anesthetic implications. We discuss successful management of a 5 years old child with infantile nephropathic cystinosis and Fanconi's syndrome who underwent osteotomy for rickets.

1. Introduction

Cystinosis is a lysosomal storage disease characterized by abnormal accumulation of amino acid cysteine. It is a genetic disorder that typically follows an autosomal recessive inheritance pattern. Prevalence of cystinosis is approximately 1:100,000 to 1:200,000.

Cystinosis affects various tissues of the body including the cornea, conjunctiva, bone marrow, lymph nodes, leukocytes, brain, muscle, intestines, thyroid, and the internal organs such as the liver, spleen, and kidneys. The latter results in Fanconi's syndrome and renal insufficiency, often necessitating the need for kidney transplant. Renal losses of calcium and phosphate may result in bony abnormalities and fracture also. We discuss successful management of a 5 years old child with infantile nephropathic cystinosis and Fanconi's syndrome who underwent osteotomy for rickets.

2. Case history

A 5 years old, 15 kg child was scheduled for osteotomy following diagnosis of rickets. The child was normal up to one year of age when he developed polyuria, severe weakness and low weight according to age. The child was operated for Hirshsprung's disease at the age of two years after which patient did not show any signs of reversal from anesthesia. On arterial blood gases, pH of the patient was found to be 7.20 and bicarbonate was 8 mmol/L. Patient was given I.V. bicarbonate and fluid after which pH became normal and patient was extubated second post-operative day. After complete workup he was diagnosed as a case of cystinosis. Child was also found to be hypothyroid and was on eltroxin 50 mcg per day supplementation. Cystiamine 200 mg qid dose, potassium, calcium, bicarbonate and phosphate supplementation also started.

Preoperative investigation revealed normal hemogram, random blood sugar was 120 mg/dl, serum creatinine and blood urea nitrogen were 0.53 mg/dl and 27 mg/dl respectively. His serum sodium was 139 mmol/L, potassium 4.2 mmol/L, ionised calcium 0.86 mmol/L, chloride 108 mmol/L and phosphate 2.2 mg/dL and serum magnesium 1.3 mEq/L. Among electrolytes, the value of ionised calcium and serum magnesium was below whereas phosphate was above the normal range. Thyroid function test and liver functions were normal. ECG and ECHO were within normal limits. On clinical examination no eye involvement or photophobia was found in this child.

Child was advised NPO for 6 h. Inj Isolyte P was started at the rate of 50 ml/hour from morning of surgery. Patient was given his usual dose of eltroxin. ABG was repeated on the day of surgery which showed serum potassium and calcium to be low. (S. K 2.8 mmol/L and ionised Ca 0.6 mmol/L). Electrolyte correction started in the preoperative area. Child was premedicated with IV

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ketamine (8 mg) and IV glycopyrrolate (60 μg) and anesthesia was induced with propofol (30 mg) and laryngeal mask airway of size 2 was inserted. Patient was given caudal block with levobupivacaine (10 ml of 0.25%) and clonidine (20 μg) after induction. No relaxant was used and child was maintained on sevoflourane on pressure support mode of ventilation. Child's intraoperative course remained uneventful. Child was given intravenous paracetamol for postoperative pain. Serum electrolytes and arterial blood gas were repeated every 6 h and were found to be normal. Patient was discharged on 3rd postoperative day.

3. Discussion

Cystinosis is caused by mutation in the CTNS genes that code for cystinosin, the lysosomal membrane specific transporter for cysteine. After the degradation of endocytosed protein to cysteine within lysosomes, it is normally transported to the cytosol. But if there is a defect in carrier protein, cysteine is accumulated in lysosome. As cysteine is highly insoluble, when its concentration in tissue lysosomes increases, its solubility is immediately exceeded and crystalline precipitates are formed in almost all organs and tissues

Three clinical patterns have been described in patients with cystinosis including the infantile or nephropathic form, an adolescent form, and an adult form. [1]. Patients with infantile nephropathic form of cystinosis develop symptoms early in life and, if left untreated, develop end stage renal failure by ten years of age [2]. Presentation often includes failure to thrive, vomiting, fever, rickets, Fanconi's syndrome with polyuria, dehydration and metabolic acidosis which were present in this patient. Crystal deposition is prominent in the eyes leading to photophobia. Hypothyroidism, insulin-dependent diabetes mellitus, and delayed puberty are often evident later in the course of illness.

In juvenile/adolescent form, ocular manifestations are predominant, renal involvement is rare and occurs in second decade of life and course is more benign than infantile form. The adult form mainly presents with ocular signs and symptoms.

As cystinosis has multiorgan involvement, it has several anesthetic implications, although the issue of primary concern is underlying renal involvement. Fanconi's syndrome is a defect in the proximal tubular transport of amino acids, proteins, glucose, phosphate, uric acid, and various electrolytes (Na^+, K^+, HCO3, phosphate)) [3]. The hypophosphatemia may lead to osteomalacia and rickets. The latter was the complaint with which child presented.

We have chosen Isolyte P for maintenance as it contains high milliequivalents of potassium and phosphate and also contains magnesium. (Sodium-23 mEq/L, potassium 20 mEq/L, Magnesium-1.5 mEq/L, Acetate-23 mEq/L, Phosphate-1.5 mEq/L, Chloride-20 mEq/L) Requirement of intravenous fluid was calculated according to weight of the patient based on holiday segar formula. Fluid status of the patient was assessed by the clinical features such as vitals and urine output. Along with this preoperative ABG was done to calculate lactate level.

We preferred propofol for induction as it has no nephrotoxic profile. Atracurium or cisatracurium should be preferred as muscle relaxants. Suxamethonium should be avoided as approximately 24% of cystinotic patients may experience a distal, vacuolar myopathy [4]. Patients with clinical myopathy may require post op mechanical ventilatory support. Neuromuscular monitoring should be used in cases where relaxant is being used. However in this case we did not use any muscle relaxant as caudal block effect was sufficient to provide effective sensory blockade. Regional anesthesia and peripheral blocks should be used cautiously as it may have legal implications. Serum electrolytes, blood sugar and ABG should be done at frequent intervals. Sevoflourane was used for maintenance of anesthesia. Studies have demonstrated no deleterious effects of sevoflurane on renal function even in patients with pre-existing renal insufficiency [5].

In conclusion, the anesthetic management of a patient with cystinosis should focus on quantifying end organ damage and methods to prevent their further deterioration during intraoperative course. Patient should continue electrolyte supplementation as well as other drugs preoperatively. ABG should be repeated at regular interval perioperatively. Nephrotoxic drugs should be avoided and all drugs should be given in titrated doses.

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