Anesthetic Management of a Patient with Pompe Disease & Kartagener Syndrome

Pompe Hastalığı & Kartagener Sendromlu Hastada Anestezi Yönetimi

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ABSTRACT

Pompe disease is an autosomal recessive disorder characterized by prominent cardiomyopathy, hepatomegaly, skeletal muscle weakness, and hypotonia. Kartagener Syndrome is a rare genetic disorder that has the triad of sinusitis, bronchiectasis, and situs inversus.
We report our experience of management of anesthesia in an infant with Pompe disease & Kartagener Syndrome who underwent general anesthesia for venous port insertion. The patient had growth retardation, severe respiratory failure, global hypertrophic left ventricle, and dextrocardia. The existence of various comorbidities and severe respiratory failure, in this case, led us to use general anesthesia with fewer medications as possible for being on the safe side of anesthesia management.
Bulbar and facial muscle weakness, lung and heart problems, multiple drug use, presence of multiple systemic clinical disorders make anesthesia management difficult in such patients. Therefore, we preferred to use dexmedetomidine, fentanyl, and low dose muscle relaxants for patient safety

Keywords: Pompe disease, Kartagener Syndrome, anesthetic management, case report

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ÖZET

Pompe hastalığı, belirgin kardiyomiyopati, hepatomegali, iskelet kası zayıflığı ve hipotoni ile karakterize otozomal resesif bir hastalıktır. Kartagener Sendromu, sinüzit, bronşektazi ve situs inversus triadına sahip nadir bir genetik bozukluktur.Bu olgu sunumunda venöz port yerleştirilmesi için genel anestezi uygulanan Pompe hastalığı ve Kartagener Sendromlu bir bebekte anestezi yönetimimizi bildiriyoruz. Hastada büyüme geriliği, ciddi solunum yetmezliği, global hipertrofik sol ventrikül ve dekstrokardi mevcuttu. Olguda, çeşitli komorbiditelerin ve ciddi solunum yetmezliğinin varlığı, anestezi yönetiminin güvenli tarafında kalmak amacı ile mümkün olduğunca az ilaç ile genel anestezi uygulamamıza neden oldu. Bulbar ve fasiyal kas güçsüzlüğü, akciğer ve kalp sorunları, birden çok ilaç kullanımı, çoklu sistemik klinik bozuklukların varlığı bu hastalarda anestezi yönetimini güçleştirmektedir. Bu nedenle, bu olgunun anestezisinde deksmedetomidin, fentanil ve düşük doz kas gevşetici kullanmayı tercih ettik.

Anahtar Sözcükler: Pompe hastalığı, Kartagener Sendromu, anestezi yönetimi, olgu sunumu

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INTRODUCTION

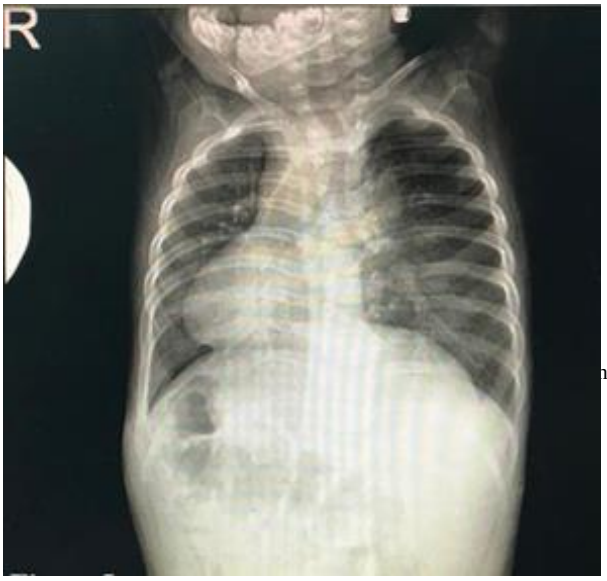
Pompe disease (PD), is an autosomal recessive disorder and is caused by mutations in the gene encoding the acid α-glucosidase (GAA) which leads to inhibition of the lysosomal degradation of glycogen, causing cell damage. The cellular pathology mainly affects skeletal, cardiac, and smooth muscle fibers. The clinical spectrum ranges from the classic form with early-onset and severe phenotype to non-classic form with later onset and milder phenotype. The classic form (infantile form) patients manifest rapidly progressive disease characterized by prominent cardiomegaly, hepatomegaly, weakness, hypotonia, and death due to cardiorespiratory failure in the first year of life (1).

Kartagener Syndrome is also an autosomal recessive disorder characterized by dysfunction of dynein arms in the cilia structure of the pulmonary system leading to the triad of sinusitis, bronchiectasis, and situs inversus (2).

Bulbar and facial muscle weakness, lung and heart problems, multiple drug use, presence of multiple systemic disorders make anesthesia management difficult in such patients. In this case report, we aim to share our experience on the management of anesthesia in an infant with PD & Kartagener Syndrome who was scheduled for venous port insertion.

CASE REPORT

Port-a-Cath (PAC) insertion planned patient with PD & Kartagener Syndrome was preoperatively evaluated for anesthesia management. 11 months old, 6800 gr, male patient who was born via spontaneous vaginal delivery at thirty-ninth weeks in an inbred family got Pompe syndrome diagnosis at the age of five months. The patient has been receiving alglucosidase α enzyme therapy biweekly since the diagnosis of the disease. Besides, there was a family history of a brother who had died at the age of eight months due to PD. Growth retardation, intercostal retractions and dextrocardia were found during physical examination. Electrocardiography was normal. Echocardiographic imaging revealed patent foramen ovale, an increase in the posterior wall and interventricular septum thickness as global hypertrophic left ventricle and dextrocardia. Situs inversus totalis was confirmed with the posteroanterior chest radiography (Figure I) and abdominal computerized tomography (Figure II). The patient's complete blood count, biochemical and coagulation parameters were within normal levels. The patient was classified as ASA IV according to American Society of Anesthesiology physical status classification system. Therefore, difficult airway and cardiopulmonary resuscitation equipment was kept ready.



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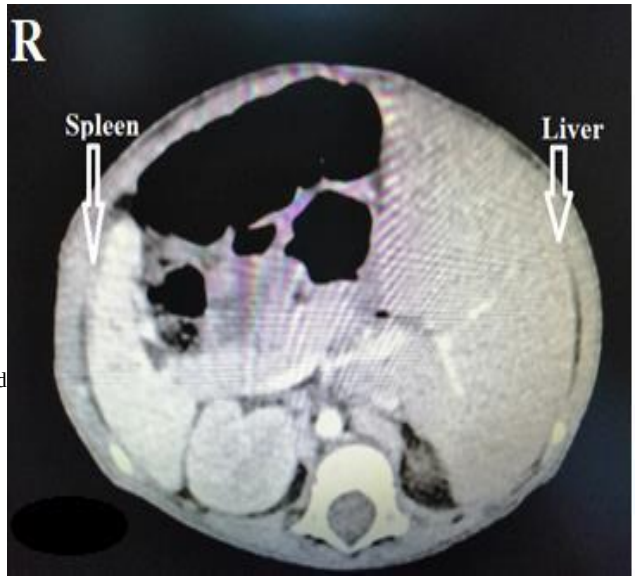

Premedication with sedative drugs was not preferred before the operation. Bispectral index (BIS) monitoring was performed in addition to standard monitoring with electrocardiogram (ECG), pulse oximeter, noninvasive blood pressure. ECG monitorization was done from the right hemithorax, the mirror image of the routine monitorization (3). Anesthesia induction was performed by 1 $\mu g.kg^{-1}$ dexmedetomidine infusion within 10 minutes followed by a maintenance dose of 0.5-0.7 $\mu g \cdot kg^{-1} \cdot hour^{-1}$. For muscle relaxation, low dose rocuronium (1.5 mg) was given in fractions and also 1 $\mu g.kg^{-1}$ fentanyl iv was administered slowly for the analgesia of the patient. After the intubation, catheterization was done through the right internal jugular vein via open surgical technique with head turned to the left 45 degrees in supine position. Operation lasted 1 hour and no problem was encountered during the operation. Dexmedetomidine was diminished stepwise and ceased at the end of the procedure. Following spontaneous extremity movement and respiration, reversal agents, neostigmine and atropine were intravenously administered. After the extubation, the patient was taken to the postoperative care unit. The patient was transferred to ward 115 minutes after the procedure with an Modified Aldrete Score of 9.

DISCUSSION

In PD and/or Kartagener Syndrome, cardiac abnormality is an expected problem. Coronary perfusion deteriorations, related to systemic vascular and diastolic arterial pressure reductions may cause malignant and highly lethal arrythmias during anesthetic management of these patients. In the series of 139 cases that Wang et al. presented, 6% of infantile Pompe patients were observed to have an arrhythmia or cardiopulmonary arrest during anesthesia induction. They emphasized that these complications occur mostly during induction with propofol and sevoflurane which have the potential to cause hypotension, as well as in patients with high left ventricular mass index (>350 g/m^2) (4).

Sevoflurane should be used in very low concentrations for induction and maintenance to avoid hypotension and cardiac arrhythmias (4). Ketamine is recommended for maintaining systemic vascular resistance and contractility without decreasing preload (5). But in a clinical pathology that leads to left ventricular outflow tract obstruction, it may increase the degree of obstruction. Propofol should be avoided during induction and maintenance as it reduces afterload, diastolic pressure, and systemic vascular resistance significantly, especially in higher doses (6). As an ideal anesthetic option, etomidate has a stable cardiovascular profile for induction in the untreated Pompe infant with severe cardiomyopathy. It has been shown that the inotropic effects of etomidate are not different in normal and cardiomyopathic hamsters (7). Based on limited data, it is predicted to be a safe agent for the patients diagnosed with PD. But, the number of studies on children is insufficient so that recommendation for its use is not supported by high-level evidence. One case with PD regarding the use of dexmedetomidine without muscle relaxants for thoracolumbar kyphoscoliosis corrective surgery was reported in the literature (8).

Considering the pathophysiological chances in PD patients, we performed the anesthesia induction with dexmedetomidine an alpha 2 agonist which provides more stable cardiac profile, especially by increasing the mean arterial pressure and systemic vascular resistance index (9).

Another reason for choosing dexmedetomidine is avoiding tachycardia which may lead to disruption of coronary perfusion.

Excessive accumulation of glycogen within the lysosomes of the respiratory muscles, especially diaphragm, results in potential problems with respiratory management. Besides, increased sensitivity to neuromuscular blockade agents (NMBAs) and opioids in patients with PD may aggrevate respiratory failure resulting from hypotonia and neuromuscular weakness. Postoperative prolonged mechanical ventilation is also a potential problem, weaning may be difficult in patients with PD. Therefore, NMBAs should be used cautiously. Suxamethonium should be avoided because of possible adverse events like hyperkalemia and rhabdomyolysis. However, no case reports of such a reaction exist in the literature. Non-depolarising NMBAs should be avoided or used only in very low doses, being concerned about increased hypotonia and weakness in these patients (10).

CONCLUSION

We believe that anesthesia management with dexmedetomidine, fentanyl, and low dose NMBA is a safe and efficient method for patients with PD &Kartagener syndrome.

Conflict of interest

No conflict of interest was declared by the authors.

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