Case Report

Anaesthetic management of a patient with Pompe disease for kyphoscoliosis correction

Address for correspondence:
Dr. Vaishali Kumbar,
Department of Anaesthesia,
Manipal Hospital, 98,
Hal Airport Road, Bengaluru,
Karnataka, India.
E-mail: drvaishalikumbar1978@gmail.com

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Vaishali Kumbar, Jayashree Simha, Parameswara Gundappa
Department of Anaesthesiology, Manipal Hospital, Bengaluru, Karnataka, India

ABSTRACT

Pompe disease (PD) is a type II glycogen storage disease, characterised by abnormal glycogen deposition, mainly in heart and skeletal muscles, leading to progressive loss of muscle function. The infantile variety is associated with severe hypertrophic cardiomyopathy and generally do not reach adulthood. The juvenile variety presents with progressive muscle weakness and respiratory failure. Anaesthetic management concerns in the patient reported here were mainly due to respiratory failure, myopathy and sensitivity to muscle relaxants and significant haemodynamic changes perioperatively. We successfully managed a 13-year-old girl with juvenile PD on respiratory support scheduled for thoracolumbar kyphoscoliosis corrective surgery. Ketamine and dexmedetomidine were used for induction of anaesthesia and maintenance. Muscle relaxants were diligently avoided in this case.

Key words: Anaesthesia, cardiomyopathy, dexmedetomidine, glycogen storage disease, kyphoscoliosis, motor evoked potentials, Pompe disease

INTRODUCTION

Pompe disease (PD) is an autosomal recessive muscular disorder of glycogen storage disease type II, caused by deficiency of lysosomal enzyme Acid Alfa-Glucosidase with an incidence of 1:40,000.[1] Infantile PD manifests within first few months of life with hypertrophic cardiomyopathy, generalised muscle weakness and hypotonia. These children die within the 1st few years of life due to progressive cardio respiratory failure or arrhythmias.[1] Childhood or juvenile PD can present at any age, and is characterised by minimum cardiac involvement, progressive skeletal muscle dysfunction with a better short-term prognosis.[1,2] Proximal lower limb and paraspinal trunk muscles are usually affected first[1] and present with kyphoscoliosis, orthopaedic deformities, muscular fibrosis and other deformities. Involvement of diaphragm and accessory muscles of respiration leads to respiratory failure. The enzyme replacement therapy (ERT) with recombinant human acid Alglucosidase Alfa has been shown to be effective in both early and late onset PD.

CASE REPORT

A 13-year-old Asian girl weighing 24 kg was admitted with progressive thoracolumbar kyphoscoliosis deformity since 2 years. She underwent tracheostomy 6 years before, related to severe bronchopneumonia management. Since then, she continued to require ventilatory support at night. Genetic study confirmed PD and she was started on ERT. Her motor milestones were delayed with progressive deterioration. At admission, she was on tracheostomy and required ventilatory support during sleep with BIPAP mode. She was not able to stand and walk.

She was conscious, oriented with normal higher mental functions. She had thoracolumbar kyphoscoliosis extending from D_1 to L_4 level. She had wasting of the lower limb and paraspinal muscles with 3/5 motor power. She had varus deformity of the left foot, bilateral fixed flexor deformity of lower limbs. Upper limb showed normal muscle power with no wasting. On auscultation, chest was clear with no

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added sounds. Oxygen saturation was 95% on room air. Blood gases were within normal limits. Chest X-ray showed normal lung fields. Her cardiovascular system and electrocardiogram (ECG) were normal. Echocardiography showed normal ventricles with ejection fraction of 54% with no evidence of hypertrophy or obstructive cardiomyopathy. Serum creatinine kinase was raised. She was prepared with chest physiotherapy and salbutamol nebulisation.

General anaesthesia was planned. No pre-medication was given. Monitoring included ECG, non-invasive blood pressure, pulse oximetry (SpO_2), end-tidal CO_2, central venous pressure, depth of anaesthesia monitoring (Entropy©), temperature and urine output. After adequate pre-oxygenation, anaesthesia was induced with dexmedetomidine infusion 1 μg/kg over 10 min, ketamine 50 mg and morphine 2 mg. Anaesthesia was maintained with dexmedetomidine infusion at 0.5 μg/kg/h, oxygen in air (50:50%) and sevoflurane 1–2% as required. The child remained haemodynamically stable. Body temperature was maintained around 36°C by active forced hot air blankets. Motor evoked potential (MEP) monitoring was used during correction of kyphoscoliosis. The total blood loss was about 1.1 L. Under central venous pressure monitoring, 450 ml of blood along with 2.3 L of Ringers lactate were infused. At the end of the surgery, child was conscious; breathing appeared shallow and hence was shifted to paediatric intensive care unit for elective post-operative ventilation. Post-operative pain was treated with fentanyl infusion.

On the 2nd post-operative day, she developed bilateral mild pleural effusion which was treated with diuretics and antibiotics. On the 5th post-operative day, patient was weaned off from ventilator. However, she continued to require nocturnal ventilatory support. Rest of the hospital stay was uneventful and she was discharged on the 10th day.

DISCUSSION

PD patients may present for correction of kyphoscoliosis and orthopaedic deformities. Respiratory failure develops early due to weakness of diaphragm and accessory muscles of respiration, decreased vital capacity and may result in chronic sleep hypoventilation, chronic CO_2 retention.[1,3] Patients with PD show increased CPK due to ongoing muscle damage.

Report of PD patients undergoing anaesthesia is rare. Most cases reported are in children undergoing muscle biopsy,[4] lens extractions.[5] Late onset PD has better life expectancy, and hence they may present for major orthopaedic corrections.[6] Dons-Sinke *et al*. reported two patients undergoing caesarean section under regional anaesthesia.[2] Extensive literature survey did not yield any report of correction of kyphoscoliosis in PD, and the present case may be the first one to be reported.

Anaesthetic concerns in this patient were related to severe myopathy, respiratory failure, blood loss and haemodynamic changes due to major surgery, hypothermia and metabolic disturbances.[7] No muscle relaxant could be used due to myopathy and also for monitoring of MEPs. Post-operative concerns were related to post-operative ventilation, lung atelectasis, pain and infections.

Ketamine increases systemic vascular resistance and contractility and less likely to reduce preload and has been found safe even in patients with hypertrophy of ventricles.[4,7,8] Hyperthermia and non-diabetic ketoacidosis have been reported in a child with PD after enflurane and succinylcholine. Even though this was transient, it raises the possibility of possible malignant hyperthermia in PD.[7] However, no report of overt malignant hyperthermia has been reported in PD. Propofol does not appear to be ideal drug for induction in PD because it causes afterload reduction and lower diastolic pressure can result in cardiac ischaemia, particularly in patients with myocardial hypertrophy.[1,8] Recently, dexmedetomidine has been used successfully in spine surgeries.[9] It provides analgesia, sedation, haemodynamic stability and decreases the requirement of other anaesthetic agents. Better perioperative haemodynamic control, no respiratory depression, analgesia, anxiolysis with no hangover effects and reduction of post-operative shivering are potentially beneficial in improving outcome.[9]

Patient developed mild bilateral pleural effusion on the 2nd post-operative day with mild respiratory difficulty. This was thought to be due to slight fluid overload and was managed with diuretics and morphine. Inspite of heart being apparently normal in our patient, it is possible that underlying mild derangement in cardiac function may still occur.

CONCLUSION

Patients with PD are often very sick. Definitive post-operative plan for ventilation and weaning from

ventilation to baseline respiratory status are key factors in managing case for major surgery. Avoiding muscle relaxants should always be considered. Both overloading and under loading of fluids should be avoided for successful outcome of the patient. Perhaps ERT helps in minimising the complications compared to overt disease.

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Conflicts of interest
There are no conflicts of interest.

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