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Case Report

A 48-Year-Old Female with Kearns-Sayre Syndrome Posted for Hysteroscopic Polypectomy

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ABSTRACT

Kearns-Sayre Syndrome (KSS) is a rare mitochondrial cytopathy caused by large deletions in mitochondrial DNA, leading to impaired energy production in cells. It has an estimated incidence of 1 to 3 cases per 100,000 population. KSS has no racial or sex predilection. The significant concerns for an anesthesiologist associated with this syndrome are multi-system involvement, including cardiac conduction defects, endocrinopathies, sensorineural deafness, and neurological dysfunction. These manifestations make it challenging for the anesthesiologist. These concerns significantly influence the choice of anesthesia given to the patient, particularly regarding the selection of neuromuscular blockers and their effects on patients with this syndrome compared to those in the general population. Anesthetic management of Kearns-Sayre syndrome patients requires careful preoperative assessment of cardiac, neurological, and metabolic status due to risks of heart block, malignant hyperthermia, respiratory muscle weakness, and lactic acidosis. Also, intraoperative and postoperative vigilance is necessary to prevent any adverse events. We report a 48-year-old female, a known case of Kearns-Sayre Syndrome, scheduled for hysteroscopic polypectomy. This case highlights the importance of detailed preoperative evaluation and tailoring the anesthetic technique to minimize mitochondrial stress.

Introduction

Kearns-Sayre syndrome (KSS) is a rare, progressive disorder with an incidence of 1 to 3 per 1,00,000 caused by defects in mitochondrial DNA [1]. KSS is defined by the triad of onset before the age of 20 years, chronic progressive external ophthalmoplegia (CPEO), & pigmentary retinopathy. Other typical clinical features of KSS include ptosis, ophthalmoplegia, cardiac conduction defects and/or cardiomyopathy, sensorineural deafness, muscle weakness, ataxia, dementia, nystagmus, developmental delay or regression, mental retardation, scoliosis, and endocrine disorders.

Affected individuals may also have one of the following conditions, such as cerebrospinal fluid (CSF) protein of more than 100 mg/dL, cerebellar ataxia, short stature, deafness, and dementia [1].

Case Report

Our patient, a 48-year-old female with an established diagnosis of Kearns-Sayre syndrome, was posted for hysteroscopic polypectomy of cervical polyps. Further evaluation revealed clinical features like progressive external ophthalmoplegia with bilateral ptosis (Figure

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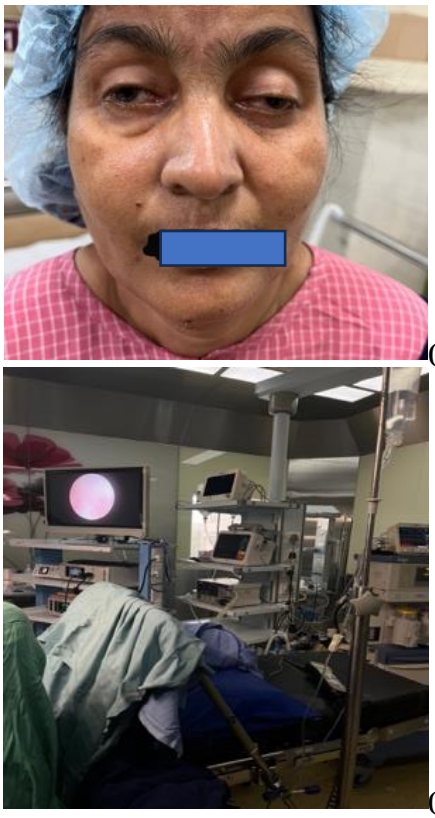
1a), exertional breathlessness, and difficulty swallowing for 2–3 years. On examination, significant findings were decreased air entry on the right side, a thyromental distance of less than 6.5 cm, a short neck suggestive of an anticipated difficult airway, and room air oxygen saturation of 93%. Advised pre operative workup, electrocardiogram (ECG) and 2D echocardiogram (2D Echo) were within acceptable limits. She showed newly diagnosed diabetes mellitus with an HbA1c of 6.6, for which she was not taking medications. Electromyography confirmed neuromuscular involvement consistent with her mitochondrial myopathy; Magnetic Resonance Imaging (MRI) of the brain showed age-related cerebral atrophy and bilateral periventricular white matter signal abnormalities representing chronic microangiopathic ischemic changes (Figure 1a).

Key risks identified were heart block due to cardiac conduction abnormalities, hypoventilation due to respiratory muscle weakness, lactic acidosis, exaggerated sensitivity to some anesthetic agents, and altered responses to neuromuscular blockers.



(a)

(b)

Figure 1- (a)Patient with clinical features of chronic progressive external ophthalmoplegia (CPEO); (b) Operation theatre during the hysteroscopy

A day before surgery, regional anesthesia was planned, and subarachnoid blockade was explained. Consent was obtained from the patient and relatives. The preoperative fasting period was minimized by keeping fasting hours of 6 hours and requesting surgeons to schedule the case as the first case of the day. Intravenous dextrose was advised to prevent hypoglycemia and catabolic stress. On the day of surgery, subarachnoid block (spinal anesthesia) was given in a sitting position under all aseptic precautions at lumbar 3-4 intravertebral space with injection of bupivacaine hydrochloride with dextrose 0.5% (ANAWIN Heavy 0.5%) 2 ml after confirming free flow of CSF. The level of block was achieved till the T10 dermatome. Intravenous fluids without lactate were used. The duration of surgery lasted for one hour, during which the patient tolerated the anesthesia and surgery well. Postoperatively, the patient was shifted to the post-anesthesia care unit (PACU) for observation. After one and a half hours, the patient was shifted after fulfilling the discharge criteria of PACU. On discharge, the patient was advised to have regular follow-up.

Discussion

KSS is a mitochondrial cytopathy often caused by large deletions in mitochondrial deoxyribonucleic acid (mtDNA), leading to defective oxidative phosphorylation in mitochondria. When oxidative phosphorylation is compromised, tissues with high energy demands like the heart, skeletal muscle (including respiratory muscles), and the brain are especially vulnerable. When mitochondrial adenosine triphosphate (ATP) production is impaired, the cells shift toward anaerobic metabolism, leading to accumulation of lactate and pyruvate and potentially lactic acidosis [2].

The phenomenon of heteroplasmy (mixture of normal and mutant mitochondria in different cells or tissues) means clinical severity varies widely, with some patients mildly affected and others severely. The various surgical and anesthetic stresses due to fasting, pain, shivering, hypotension, hypoxia, and hypovolemia may push the patient from a compensated to a decompensated metabolic state [3]. Intracranial degenerations are common, and elevated protein in the CSF indicates damage to the central nervous system. Sometimes mitochondrial myopathies can mimic myasthenia gravis, as proximal muscle weakness and ptosis are the main presenting symptoms [4]. KSS involves multiple organs, and the symptoms primarily arise in childhood [5]. Respiratory dysfunction may develop in association with suppression of ventilatory drive to hypoxemia and hypercapnia, sensitivity to anesthetic induction agents, and the prolonged effect of neuromuscular blocking agents [6]. Prolonged periods of nil by mouth and use of Ringer's lactate for hydration should be avoided to prevent lactic acidosis. Instead, use of glucose-containing solutions may help in preventing anaerobic metabolism and lactic acid production [7].

Pre-medication with sedatives and opioids is avoided because the respiratory response to hypoxemia is impaired. If general anesthesia (GA) is given, then weaning from mechanical ventilation could be difficult, presenting as recurrent pneumonia and respiratory failure. Sensitivity to volatile agents, barbiturates, propofol, and etomidate has been suggested in patients with mitochondrial disease. Cardiostable agents should be used, as these patients are prone to conduction abnormalities. Propofol can also disrupt the mitochondrial permeability transition pore, leading to a decrease in mitochondrial membrane potential and subsequent apoptosis. Additionally, the inhibition of free fatty acid uptake into the mitochondria may be a causal factor for propofol infusion syndrome [8-9].

Patients with KSS require a cautious plan of anesthesia to minimize drug exposure and hemodynamic instability. Subarachnoid block offers excellent operating conditions in the lower abdomen and lower extremities with the least amount of drug transfer. It also avoids any complications arising from the use of neuromuscular blockade in general anesthesia, like residual neuromuscular paralysis or the side effects arising from the use of opioids and induction agents. These patients can have autonomic neuropathy, which impairs their cardiovascular system's ability to compensate for blood pressure changes. Even the exaggerated and prolonged drop in blood pressure of the sympathetic blockade of spinal anesthesia may be difficult to treat with standard vasopressors. Hence, the use of a smaller concentration of local anesthetic for such procedures in KSS seems prudent in preventing the incidence of hypotension [7,10].

Conclusion

Patients with KSS require tailored anesthetic planning, taking into account their mitochondrial dysfunction. Understanding mitochondrial bioenergetics helps anticipate perioperative risks: lactic acidosis, muscle weakness, sensitivity to drugs, and cardiac conduction problems.

Regional techniques are advantageous over general anesthesia, but if required, GA must be carefully tuned. Choice and dosage of agents are crucial. Prevention of metabolic derangement should be taken care of by avoiding hypoglycemia, unnecessary lactate load, hypoxia, hypotension, and shivering. Close perioperative monitoring and readiness to manage the conduction, ventilation, and metabolic issues are critical. The number of reports on anesthesia management of KSS are few. Our case report attempts to put forth a concise and comprehensive approach for this rare syndrome for all the anesthesiologists who may encounter it in the future.

Acknowledgment

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