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CASE REPORT

Sanjad–Sakati Syndrome: An Anesthetic Challenge

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Abstract

Due to scarce data regarding "Middle Eastern Syndrome" or the Sanjad–Sakati syndrome, this case report aims to elaborate the significance of this rare autosomal recessive syndrome and the considerations to provide anesthesia to such patients. The patient discussed in this report is a diagnosed case of Sanjad–Sakati syndrome, indicated for surgical insertion of gastrostomy tube by laparotomy. The anesthetic concerns encountered included a difficult airway anatomy due to facial dysmorphic features and short stature, along with increased sensitivity to muscle relaxants. The case was further complicated by the presence of chest infection and electrolyte imbalance, particularly hypocalcemia, with the risk of seizures, cardiovascular collapse, and arrhythmias. A thorough preoperative assessment and multidisciplinary approach, including correction of electrolytes and optimization of the chest condition, followed by a cautious and vigilant intraoperative anesthetic management and close postoperative monitoring led to a smooth recovery and positive outcome with the avoidance of any morbidity or mortality.

Keywords: Anesthesia, difficult airway, hypocalcemia, hypoparathyroidism-retardation-dysmorphism, Sanjad–Sakati syndrome

Introduction

Sanjad-Sakati Syndrome is a rare autosomal recessive genetic disorder. It is caused by mutations in the gene-encoding tubulin-specific chaperone E (TBCE), located on chromosome 1q42.3.[1] The syndrome was first identified and reported in the pediatric population of the Kingdom of Saudi Arabia during 1988.[2] Since the cases that were further reported were confined to the regions of Arabian Peninsula and North Africa, it is thought to be associated with consanguinity, and hence, also came to be popularly known as the Middle Eastern syndrome.[3]

Another synonym for the syndrome is hypoparathyroidism-retardation-dysmorphism (HRD) syndrome. The affected child is usually born with intrauterine growth restriction (IUGR) complicated with tetany and seizures due to the associated characteristic hypocalcemia, hyperphosphatemia, and hypoparathyroidism. These patients are usually physically and mentally retarded with typical physical findings inclusive of dwarfism, micrognathia, large ears, thin long fingers, and dentition abnormalities.[4, 5]

Case report

A 13-year-old girl was referred to the department of Anesthesia for laparotomy and feeding gastrostomy procedure. She had been diagnosed to have Sanjad–Sakati syndrome, along with α thalassemia trait, sickle cell trait, hypothyroidism, and glucose-6-phosphate dehydrogenase (G6PD)

decreased activity. Preoperative assessment and evaluation of the patient revealed weight as 7 kg with typical features of Sanjad–Sakati syndrome and a positive family history of the syndrome. She was born to consanguineous parents, full term, by normal vaginal delivery, birth weight of 2.6 kg, and history of multiple hospital admissions postnatally for seizures, electrolyte imbalance, and recurrent chest infections. Further, she developed complications, such as nephrocalcinosis and calcification deposits in the cornea and the brain. Patient had previous history of general anesthesia for dental surgery, which was performed at the age of 8 years.

Preoperative laboratory investigations included abnormal levels of calcium (1.97 mmol/l), phosphorus (1.9 mmol/l), and thyroid stimulating hormone (0.01 mU/L) which were corrected. The remaining parameters, such as the complete blood count and the coagulation profile were within normal limits. Ongoing medications of the patient included calcium, vitamin D_3, and potassium supplements along with L-Thyroxine 50µg once daily. These regular medications were continued even on the day of the surgery according to her routine dosing schedule.

Intraoperatively, as difficult airway was anticipated, the difficult airway trolley was kept standby, and patient was induced with sevoflurane (8%) and oxygen (100%) in the presence of an ENT surgeon for the possible need of an emergency tracheostomy. A prior informed consent was taken from the parents before proceeding, ensuring that it meets the ethical standards of the hospital and the medical committee. Peripheral intravenous access was obtained while maintaining spontaneous respiration. An attempt at direct laryngoscopy was performed to visualize the vocal cords and the trachea was successfully intubated with the help of a stylet and external laryngeal manipulation, following which 2 µg/kg fentanyl was administered. The use of muscle relaxants was avoided. Anesthesia was maintained with bolus doses of propofol (10 mg) and fentanyl (5 µg) along with oxygen (50%), air (50%), and sevoflurane (3%). Patient remained vitally stable throughout the procedure and extubated fully awake. Recovery and transfer to the post anesthesia care unit (PACU) was eventful.

Following PACU, the patient was then transferred to the pediatric intensive care unit (PICU) for overnight monitoring of her vital signs and maintenance of oxygen saturation along with observation of development of any postoperative complications. She was then discharged to the general ward after 24 hours.

Discussion

Sanjad–Sakati syndrome is rare and to manage a patient affected with this syndrome is difficult, and hence, much needs to be still known about the disease and its implications on anesthesia. Due to the isolation of the cases to the Arabian and the Middle Eastern regions, consanguineous marriages are considered as one of the major predisposing factors.[2, 3]

The affected patients are typically born with growth and mental retardation along with the characteristic findings of dysmorphic features, such as microcephaly, elongated face, depressed nasal bridge, pointed nose, thin lips, and mandibular hypoplasia or micrognathia.[4, 5] The syndrome is usually symptomatic from early course of life[4, 5] due to seizures and tetany caused by the associated hypocalcemia, hyperphosphatemia, and congenital hypoparathyroidism.[6] Recurrent chest infections are also common, requiring frequent hospital admissions. Suffering from abnormal dentition, poor oral hygiene, recurrent fractures, and long-term need for enteral nutrition support, these patients undergo multiple surgeries for the same. Hence, it is important to understand the anesthetic considerations in such patients and outline the pre-, intra-, and postoperative concerns. To our knowledge there is no previously published study available so far, in this view.

Preoperatively, in addition to a general anesthetic history and examination, the identification of a potential difficult airway is of prime importance and the degree of dysmorphism should be established. The electrolyte and hormonal imbalances should be corrected as uncorrected low calcium levels may result in severe hypotension, laryngospasm, bronchospasm, coagulation disorders, cardiovascular collapse, cardiac dysrhythmias, and seizures.[7] In view of recurrent chest infections, chest

X-ray, arterial blood gas analysis, and pulmonary function tests must be performed. Infections thyroid and parathyroid hormone imbalance should be treated, and the chest condition should be optimized prior to the surgical procedure. If patients are known cases of epilepsy, as seizures are common in this group of patients, careful evaluation of the drugs with neurological consultation is advisable as their chronic use may alter the metabolism of drugs administered. Due to the electrolyte abnormality, these patients also suffer from psychiatric and social issues, depression, irritability, and anxiety. These group of patients may benefit from premedication with a sedative, such as a benzodiazepine. Opioids are better avoided as these patients are prone to respiratory depression and apnea. Prophylactic antibiotic coverage may be required to prevent the risk of chest infection.

Intraoperatively, such patients present as a challenge to the anesthetist. Complete monitoring, including heart rate, electrocardiography, pulse oximetry, blood pressure, end-tidal carbon-dioxide ($ETCO_2$), and temperature are essential prior to surgery. As there is a potential for difficult airway, careful clinical assessment, planning, and team work are the key to anesthetic safety and success. It is advisable that spontaneous ventilation should be maintained during intubation to avoid hyperventilation as it can precipitate hypocalcemia as well as decrease the threshold for seizures. Hence, the levels of $ETCO_2$ should be maintained around 40 mmHg. Tube selection may be done according to the child's weight rather than the age. It is important to avoid muscle relaxants, as the action of the muscle relaxants may be prolonged due to hypocalcemia.[8] However, if required, small doses may be used, in which case it is highly desirable to use neuromuscular monitoring. It is also vital to consider intraoperative positioning as these patients are very frail and are prone to fractures, especially, long bone fractures. Vigilance in monitoring is very important as use of intravenous anesthetics, such as barbiturates may cause profound hypotension due to smooth muscle relaxation. Use of volatile agents may lead to bradycardia, dysrhythmias, ST prolongation, or T wave inversion in the presence of hypocalcemia. Use of local anesthetics, especially Bupivacaine, must be cautious as risk of cardiotoxicity may be increased in the presence of hypocalcemia. Massive blood transfusion and hypothermia should be avoided as clearance of citrate may be decreased from the blood.[9] Measurement of electrolytes intraoperatively in long procedures is advisable to correct the blood or electrolyte abnormalities.

Management of calcium is critical, as serum ionized calcium level <0.5 mmol/L may lead to life-threatening complications. It is a medical emergency that requires intravenous calcium therapy consisting of correction of any existing respiratory or metabolic alkalosis and administering a calcium bolus (100–200 mg of elemental calcium over 10 mins.), followed by a maintenance infusion of 1–2 mg/kg/h of calcium. The serum calcium level returns to normal within 6–12 hours with this regimen. Thereafter, the maintenance rate can be decreased to 0.3–0.5 mg/kg/h.[7] We advise a complete awake extubation with evidence of good muscle tone and hemodynamic stability.

Postoperatively, these patients should be highly monitored in the PICU as there are reported cases of hypoventilation.[10] As it is vital to avoid the factors that are responsible for altering calcium levels, it is also important to assess the signs of hypocalcemia, such as laryngospasm, hypotension, cardiac arrhythmias, and seizures. Effective pain control is important and a multimodal approach, including nonopioid analgesics (paracetamol). Local or regional anesthesia is suggested to limit peri- and postoperative opioid requirements and to avoid respiratory depression.[11, 12]

Conclusion

Due to the rarity of Sanjad–Sakati syndrome with its prevalence in the Arabian Peninsula, it is important to provide proper genetic counselling to the families of this region for the early recognition of the disease. At the same time, it is essential for the anesthetists to perceive knowledge about the syndrome, its anesthetic implications, the challenges it may pose. The pre-, intra-, and postoperative implications require timely recognition, optimization, and skillful management in order to avoid complications. Appropriate knowledge and a multidisciplinary approach yield a successful outcome.

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