Case report

Anesthetic Management of a Four-Year-Old Child Affected by Marinesco-Sjogren Syndrome Scheduled for Cataract Surgery: A Case Report

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Abstract

Marinesco-Sjogren syndrome (MSS) is a rare autosomal recessive disorder with different clinical signs and symptoms. Neurological dysfunction, especially in the cerebellum, ataxia, dysarthria, muscle atrophy, progressive hypotonia, skeletal deformity with scoliosis, mental retardation, and failure to thrive are common. Congenital cataracts and strabismus may need to be corrected via surgery. This disorder requires unique anesthetic consideration for myopathy, malignant hyperthermia, and respiratory depression. The neurodevelopmental delay also makes anesthetic management challenging in these patients. Muscle weakness and poor respiratory reserve in these patients may be associated with adverse effects if muscle relaxants are used during anesthesia. Case reports of anesthetic management of this syndrome are scarce. We report a child with MSS scheduled for lens removal surgery and anesthetic management.

Keywords: Cataract, Cerebellar ataxia, Spinocerebellar degenerations, Muscular diseases

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Introduction

Marinesco-Sjogren syndrome (MSS) is a recessively inherited multisystem disorder with various signs and symptoms. The most important characteristics of MSS are cerebellar atrophy, ataxia, skeletal muscle deformity, short stature, progressive myopathy, inability to walk, and variable degrees of mental and sexual developmental delay. Other probable features include strabismus, nystagmus, dysarthria, and congenital cataract (1).

This rare autosomal recessive genetic disorder was first described by Marinescu et al. in 1931. SIL1 gene mutation linked to chromosome 5q31 was seen in %60 of patients (2). This gene encodes nuclear exchange factors for the endoplasmic reticulum (ER), the only gene associated with MSS (3). Although different physiopathological hypotheses were proposed for this syndrome, including chylomicron metabolism defect and lysosomal storage disorder, the primary molecular defect remains unknown. One subtype of MSS with acute rhabdomyolysis without underlying chronic myopathy was linked to the congenital cataract, facial dysmorphism and peripheral

neuropathy (CCFDN) locus, suggesting genetic homogeneity between these two syndromes. CCFDN is localized to 18qter (4, 5). There are only more than 100 patients worldwide with this syndrome. We present a child diagnosed with MSS scheduled for cataract surgery and her anesthetic management.

Case Report

The patient was a 4-year-old girl weighing 10 kg diagnosed with MSS and scheduled for cataract surgery. She was born in a second-degree consanguineous marriage and had a similarly affected older brother. Her birth history was uneventful. She had an intellectual disorder, failure to thrive, and mental and speech problems. She was unable to walk but could crawl and had four atrophic limbs. Echocardiography revealed normal findings, and there was not any history of dyspnea or shortness of breath. There was no swallowing disorder or aspiration history.

Moreover, she did not have a history of hospital admission. The brain's magnetic resonance imaging revealed severe atrophic changes at the superior aspect of the cerebellar hemispheres and vermis. Airway examinations were normal, including the open mouth, thyromental distance, head extension, and Mallampati score. There was no spine deformity or scoliosis. The primary anesthetic concerns include neurological manifestations (hypotonia and myopathy), risk of malignant hyperthermia, respiratory depression, and altered response to muscle relaxants.

After washing out the breathing circle with 100% high-flow oxygen for 10 minutes and securing an intravenous (IV) line, the patient was monitored using electrocardiography (ECG), SPO2, bispectral index (BIS) monitoring, blood pressure (BP) and ETCO2.

Atropine 0.3 mg (0.03 mg/Kg) and Fentanyl 20 microgram (2 µg/Kg) were administered, and anesthesia was induced using titrated propofol up to 30 mg (3 mg/kg). We inserted a laryngeal mask airway (LMA size 2.5) at a BIS value of 40, and the patient was ventilated with 100% oxygen using volume control mode. The anesthesia was maintained using propofol (100-200 µg/Kg/min) and remifentanil (0.1-0.2 µg/Kg) infusion. The procedure lasted two hours. Anesthesia infusion was done to keep BIS at 40-45. Vital signs and ETCO2 were normal during the surgery. At the end of the surgery and 10 minutes after stopping the anesthesia infusion, the child opened her eyes. She had reasonable respiratory effort and required no ventilator support. She was observed overnight in the pediatric intensive care unit and discharged home one day later without any event.

Discussion

MSS is a rare disorder, and anesthetic management of these patients is challenging. The disease characteristics are myopathy, mental retardation, and cerebellar ataxia. The features are usually evident at birth because of hypotonia. Treatment for MSS is only symptomatic and supportive. Therefore, there is no specific treatment for individuals with MSS. The cataracted eye lens is one of the usual features that occurs soon after birth and must be removed due to visual impairment (1). Considerations of this disease are related to neurological disorders, movement disability, hypotonia, and myopathy. Other concerns are the risk of malignant hyperthermia, respiratory depression with minimal muscular reserve, and risk of aspiration. Scoliosis and spine deformity can also cause a decrease in lung capacity and difficult intubation. In these patients, local anesthesia, regional anesthesia, or neuraxial blockade with IV sedation, if possible, are preferred over general anesthesia to prevent malignant hyperthermia and respiratory depression in the post-operation period (1). However, general anesthesia is necessary for surgeries like a cataract in mentally retarded individuals or children.

A report described a prolonged recovery period following the administration of muscle relaxants for intubation in a patient with MSS (6). Another study reported laryngeal edema necessitating tracheostomy in a child affected by MSS undergoing cataract surgery (7). Variable responses to non-depolarizing muscle relaxants in patients with myopathy are known anesthetic concerns (2).

Our case was undergoing surgery in a center with facilities for postoperative ventilation and a pediatric intensive care unit. However, we preferred to

avoid neuromuscular paralysis. The LMA provided proper ventilation, so we did not need to intubate the patient. Intubation after topical anesthesia with propofol and remifentanil instead of muscle relaxants was reserved as a backup if required. We used titrated dose of hypnotic for this patient based on BIS, so we had a proper level of anesthesia without any movement. If any muscle relaxants were needed, it was better to choose mivacurium or rocuronium because of their shorter duration of action. We also used remifentanil infusion as a short-acting analgesic. Exposure to propofol was limited to a short duration, and it was carefully titrated based on BIS monitoring because of probable mitochondrial myopathy. At the end of the procedure, the patient was awake with the same muscle tone as before, with spontaneous ventilation and normal saturation without any problem.

Conclusion

Some anesthesia considerations for patients affected with MSS include myopathy, hypotonicity, malignant hyperthermia, and delayed recovery. Regional and neuraxial anesthesia is preferred to avoid respiratory depression. Response to muscle relaxants is unpredictable, and avoiding muscle relaxants is better. Cautious monitoring and anesthetic drug titration will be helpful for safe anesthesia in these patients.

Acknowledgment

None.

Conflicts of Interest

The authors declare that there are no conflicts of interest.

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