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Bilateral Facoemulsification and Intraocular Lens Positioning in a Child with Merosin-Positive Congenital Muscular Dystrophy

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Abstract

Background: Merosin positive congenital muscular dystrophy poses significant challenges for anesthesiologists due to its low prevalence and limited research available. Additionally, there is a risk of complications arising during surgical procedures. Given the scarcity of literature on anesthetic management in patients with this condition, it becomes crucial to share previous successful experiences in handling such cases. *Case Report*: We present the case of a 6-year-old child diagnosed with merosin-positive congenital muscular dystrophy who underwent bilateral congenital cataract correction under general anesthesia. In this particular case, neuromuscular blockade was not employed, and instead, bilateral ocular peribulbar block was administered. *Conclusion*: This article aims to provide an overview of the anesthetic management of a child with a rare congenital muscular disease, highlighting the potential risks of peri-operative complications such as cardiorespiratory failure, rhabdomyolysis, and hyperkalemia.

Keywords: Anesthesia, Cataract, Congenital Muscular Dystrophy, Pediatrics.

Introduction

Merosin-positive congenital muscular dystrophy is a rare genetic disorder characterized by muscle weakness, joint contractures, and delayed motor development [1]. It presents unique challenges in the field of ophthalmology, particularly when surgical interventions such as bilateral facoemulsification and intraocular lens positioning are required in affected children. This article aims to provide a comprehensive overview of the considerations, challenges, and outcomes associated with these procedures in children with merosin-positive congenital muscular dystrophy.

Merosin-positive congenital muscular dystrophy is categorized as a subtype of congenital muscular dystrophy, characterized by mutations in the LAMA2 gene that encodes merosin, a protein crucial for basement membrane integrity [2]. This genetic defect results in abnormal muscle fiber formation, leading to progressive muscle weakness and functional limitations. In addition to muscle involvement, merosin-positive congenital muscular dystrophy can also affect other systems, including the central nervous system and cardiac function [2].

When a child with merosin-positive congenital muscular dystrophy requires bilateral facoemulsification and intraocular lens positioning, careful pre-operative assessment and planning are essential. These procedures involve delicate manipulations of the eye, necessitating an understanding of the potential challenges associated with the underlying muscular weakness, joint contractures, and associated co-morbidities. Anesthesia management is a critical aspect of these procedures. Considering the potential risks associated with muscle weakness and respiratory

compromise, a thorough evaluation of the patient's pulmonary function, airway management, and hemodynamic stability is imperative. Anesthesia techniques that minimize the use of neuromuscular blockade while ensuring patient comfort and safety should be employed. Surgical techniques must be tailored to accommodate the unique needs of children with merosin-positive congenital muscular dystrophy. The surgeon should be mindful of the potential difficulties in patient positioning, maintaining surgical field stability, and ensuring adequate exposure during the procedures. Intraocular lens selection and placement should be carefully considered, taking into account the patient's visual needs, ocular abnormalities, and potential challenges associated with the underlying muscular weakness. Post-operative care and follow-up are crucial in optimizing outcomes and managing potential complications. Close monitoring of visual acuity, ocular alignment, and post-operative healing is essential, while rehabilitation programs may be necessary to address any residual visual impairments or functional limitations.

By providing insights into the challenges, considerations, and outcomes of bilateral facoemulsification and intraocular lens positioning in children with merosin-positive congenital muscular dystrophy, this article aims to contribute to the understanding and management of these complex cases.

Case Report

A 6-year-old male child, 17.5 kg, diagnosed with a merosin-positive congenital muscular dystrophy, congenital cataract, central nervous system abnormalities (hypoplasia of the pontin, cerebellum, callous body and frontal lobes and agenesis of the pellucid septum) and myoclonic seizures presented for the correction of the congenital cataract. Neurodevelopment was overdue, important cognitive deficit was present as well as short stature, microcephalia, nystagmus, hypertonia, hyperreflexia and anterior subluxation of the hips. The patient continually received risperidone 1 mg at night and valproic acid 250 mg every twelve hours. No changes in cardiac function were observed on echocardiography. An electroneuromyography showed an electric silence when the muscle was resting and, when stimulated, the muscle fibers were intermittently recruited; many potentials had reduced amplitude and duration. A muscular biopsy showed abnormalities compatible with congenital muscular dystrophy: there was, mainly, modifications in the sarcolemmal membrane, with wrinkling and interruptions and a gap between the sarcolemma and the contractile fibers. Creatine kinase levels were higher, already expected in the congenital muscular dystrophies. No difficult airway predictors were present. In the pre-operative period, a complete evaluation of the patient was carried out together with the genetic, neuropediatric, cardiology and ophthalmology teams. The free and informed consent form was obtained for the anesthetic procedure as well as for the publication of this case report. Before the surgery, the operating theater was prepared for this patient: sevoflurane vaporizer was removed, the ventilator was washed with high oxygen flow for two hours. The anesthetic team performed a total intravenous anesthesia. In the intra-operative period, the patient was monitored with cardioscopy, pulse oximetry, non-invasive blood pressure, core temperature and end tidal carbon dioxide. A peripheral venous access 22G was punctured in the left upper limb. Propofol was administered and a laryngeal mask size 2.5 was placed. Propofol infusion was maintained at a 0.3 µg/kg/min. No neuromuscular blockade was administered during the procedure and spontaneous ventilatory drive was kept. A peribulbar block was performed in the inferolateral region of each eye utilizing a combination of 200 IU hyaluronidase, 80 mg lidocaine and 30 mg bupivacaine. Prophylaxis against nausea and vomiting was administered using 3 mg of ondansetron and 4 mg of dexamethasone. The bilateral phacoemulsification procedure and intraocular lens placement were carried out without

any complications. The patient recovered in the ICU bed and was subsequently discharged home after a two-day stay, with no reported complications.

Discussion

Patients with merosin-positive congenital muscular dystrophy who require surgical procedures, including bilateral facoemulsification and intraocular lens positioning, present unique challenges during anesthetic management [3]. Peri-operative risk assessment in these individuals can be aided by using the muscular impairment rating scale (MIRS), which categorizes patients into five grades based on the severity of muscular involvement [4]. Pre-operative evaluation should include a comprehensive cardiological assessment due to the potential presence of cardiomyopathy and arrhythmias. Additionally, predictors of difficult airway and respiratory compromise should be evaluated in this population. Patients with forced vital capacity (FVC) lower than 50% may require post-operative non-invasive ventilation, and bronchoaspiration prophylaxis should be considered [4].

Merosin-positive congenital muscular dystrophy patients undergoing surgery are at an increased risk of anesthetic-related complications, including heightened susceptibility to anesthetic agents, inherent muscle weakness, cardiorespiratory depression, and the potential for rhabdomyolysis and hyperkalemia [5]. While there is some suggestion of an association between certain muscular dystrophies and malignant hyperthermia (MH), the genetic link remains uncertain [6]. Therefore, the use of volatile anesthetics and succinylcholine should be approached cautiously, as they may trigger life-threatening rhabdomyolysis and hyperkalemia due to the breakdown of already compromised muscle membranes [5]. Total venous anesthesia may be a safer option for these patients, and if hyperkalemia occurs, treatment options include hyperventilation, intravenous insulin, sodium bicarbonate, and adrenaline administration, along with hydration and mannitol for managing high creatine kinase levels [5].

Opioids are considered safe for analgesia; however, patients with congenital muscular dystrophy are more susceptible to side effects such as respiratory depression, gastrointestinal paresis, and aspiration [5]. In this case report, an opioid-free technique was used, and analgesia was provided through an ocular block. When non-depolarizing muscle relaxants are utilized, neuromuscular monitoring becomes essential to minimize the risk of residual paralysis, which is more common in these patients [5]. It is crucial to administer fluids free of potassium, and potassium levels should be monitored during the procedure [5]. Given the higher risk of post-operative apnea and death, patients with merosin-positive congenital muscular dystrophy should recover in the intensive care unit [7].

Conclusion

The anesthetic management of patients with merosin-positive congenital muscular dystrophy necessitates consideration of their unique characteristics and challenges. A multidisciplinary approach to peri-operative care is crucial, as these patients can be complex to manage. This case report demonstrates a successful anesthetic procedure in a child with merosin-positive congenital muscular dystrophy and aims to provide valuable insights for physicians caring for patients with this extremely rare condition. By sharing our experience, we hope to improve the care and safety of individuals with merosin-positive congenital muscular dystrophy undergoing surgical interventions.

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