Case Report

Multiple pterygium syndrome: Challenge for anesthesiologist

ABSTRACT

Multiple pterygium syndrome (MPS) is a very rare autosomal recessive disorder characterized by flexion of joint and digit contractures, skin webbing, cleft palate, deformity of the spine, and cervical spine fusion. Difficult airway is associated mainly due to micrognathia, retrognathia, webbing of the neck, and limitation of the mouth opening and neck extension. We are reporting a case of a 5-year-old female diagnosed with MPS and exhibiting a bilateral club foot and congenital vertical talus. The patient was posted for manipulation and above the knee casting under general anesthesia.

Key words: Anesthesia; challenges; multiple pterygium syndrome

Introduction

Multiple pterygium syndrome (MPS) is a very rare autosomal recessive disorder. Its prevalence is not known. These patients have contracture bands around joints, and subsequently their mobility is reduced significantly. Other associated anomalies include cleft palate, syngnathia (congenital bands of tissue between the maxilla and mandible), ankyloglossia (extensive adhesions of the tongue to the palate), micrognathia, and webbing of the neck.[1] These anomalies restrict mouth opening and prevent neck extension. The tongue is displaced posteriorly, limiting the visualization of the pharynx. Many other congenital abnormalities are also associated with this syndrome. In this case report, we are discussing the anesthetic management of this rare disease MPS.

Case Report

A 5-year-old female, weighing 13 kg, diagnosed as a case of MPS, posted for manipulation and above the knee casting for a bilateral (b/l) club foot and congenital vertical talus [Figure 1]. Preoperative examination revealed multiple joint flexion contractures. Other manifestations of the syndrome include short neck with webbing, micrognathia, ankyloglossia, pectus

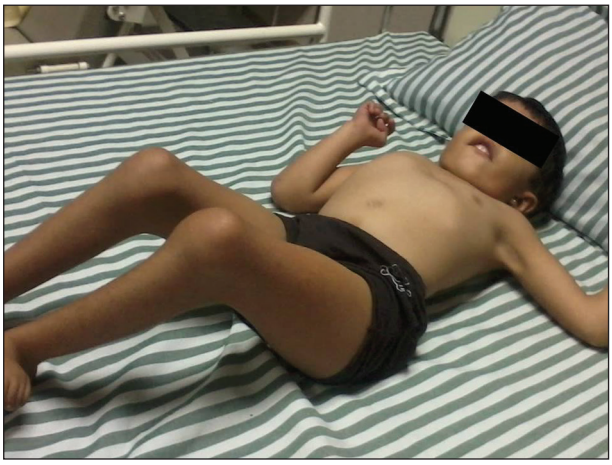

Figure 1: Child with multiple pterygium syndrome showing multiple contractures

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excavatum with right side bulge in thorax, b/l planovalgus deformity, b/l club foot since birth, b/l restricted supination with finger stiffness along with pterygia across the elbows, axilla, and popliteal fossae were also present. On examination, the cardiovascular and respiratory systems were normal. In the preoperative evaluation, heart rate was 98 beats/min, respiratory rate was 17 breaths/min, and blood pressure was 104/69 mmHg. Her neck extension was restricted (<20°) due to flexion contracture over the neck. MPG grade could not be assessed due to noncooperation of patient, but high arched palate was visible. Mouth opening was two finger breadths. Electrocardiogram (ECG) and Echocardiography were normal. The patient was scheduled to receive general anesthesia. As there were no adhesions in the oral cavity, laryngeal mask airway (LMA) insertion was planned. On the morning, the patient was shifted to operating theatre (OT) without any premedication for surgery. Intravenous (IV) cannulation was obtained in the preoperative area with a 22 gauge cannula in the right upper limb. Difficult airway cart was kept ready. In the OT, ECG, $SPO_{2,}$ and noninvasive blood pressure monitoring were started. After confirming the adequacy of mask ventilation, induction was done with IV fentanyl 25 g, IV propofol 25 mg. A size 2 LMA was inserted without difficulty, secured in place and confirmed with capnogram. Anesthesia was maintained with sevoflurane 2-3%. At the end of the procedure, sevoflurane was stopped, and LMA was removed on the recovery of consciousness and reflexes. The patient was monitored in the postanesthesia care unit for 30 min and then shifted to the ward. Intraoperative and postoperative period were uneventful.

Discussion

The clinical findings in our patient were consistent with the diagnosis of MPS (growth retardation, craniofacial dysmorphism, multiple pterygia, vertical talus, and multiple joint contractures). If left untreated, it can cause pain and morbidity which affect the patient's ambulation and quality of life.

The syndrome was first described by Matolcsy[2] and later its wide phenotypic spectrum was discussed in various reports.[3] There are two different forms of MPS, which are differentiated by their clinical severity. Lethal MPS is typically fatal in the second or third trimester of pregnancy. If delivered, they are usually stillborn or die in the early neonatal period. Pulmonary hypoplasia is considered to be the most common primary cause of mortality. MPS Escobar type (also called as Escobar syndrome) is the milder form. In patients with Escobar syndrome, webbing typically affects the skin of the neck, fingers, forearms, inner thighs, and back of the knee. These patients usually have distinct facial features as droopy eyelids (ptosis), down-slanting palpebral fissures, epicanthal folds, cryptorchidism, micrognathia, and low-set ears. These patients may also have arthrogryposis and respiratory distress at birth due to lung hypoplasia, which were not present in the case presented. These patients do not have any muscle weakness later in life. The underlying etiology is unknown. In some patients, biopsy shows the presence of muscle degeneration and disorganization of myofibrils. The most common mutation in nonlethal MPS is in the CHRNG gene, which involved in the formation of the gamma (γ) subunit of the acetylcholine receptor (AChR). The lethal form may also be associated with this gene mutation. This AChR is essential for signaling between nerve and muscle cells, which is necessary for the movement, so its mutation causes fetal akinesia resulting in pterygium (webbing) formation and congenital contractures of joints.[4] The patients may present frequently to the operating room for various surgeries as repair of cleft palate, syndactyly, scoliosis, pes equinovarus, umbilical or an inguinal hernia, and congenital hip dislocation.

Robinson *et al*.[5] reported a case of Escobar syndrome complicated with malignant hyperthermia (MH). Gericke[6] also tried to prove this association, but the current available information does not prove the relation in MPS and MH.

Escobar syndrome is a challenging condition for the anesthesiologist. Due to multiple contractures, IV cannulation and positioning may be difficult in these patients. During the preoperative period, a proper clinical evaluation should be done. An important aspect of anesthetic management in these patients includes management of difficult pediatric airway. During a preoperative check-up, factors for difficult airway as micrognathia, ankyloglossia, restriction in mouth opening, and webbing of neck and cleft palate should be looked to decide for a plan for an optimal airway management plan. These anomalies restrict mouth opening and prevent neck extension. The tongue is often displaced posteriorly, limiting the visualization of the pharynx. This syndrome is progressive. Airway management of these patients became difficult as the child grows older due to increased deformity of the airway by the pterygia and contractures. This results from increased rotation, flexion, and decreased mobility of the head and neck caused by the pterygia as the children grow larger. Positioning for laryngoscopy and glottic visualization become difficult. Airway management in most of other congenital syndrome becomes easier as the children grow older and larger, which is contrary to MPS.

Difficult airway cart should be kept ready for these patients. Preparations for alternative techniques of intubation as fiberoptic intubation (FOI), LMA, and FOI with LMA should

be made before anesthetic induction. Prior consultation with a pediatric otolaryngologist is to be considered.

Kuzma *et al*.[7] reported a case in which he initially tried awake fiberoptic-guided intubation, which was unsuccessful, followed by LMA assisted fiberoptic-guided intubation for a child with Escobar syndrome. Other alternative techniques successfully used for intubation in pediatric patients with craniofacial anomalies are video laryngoscopy, airtraq optical laryngoscope, and glide scope.[8] Mathew *et al*.[9] also used proseal LMA for ventilation followed by intubation in such patient.

In our case, mouth opening was two finger breadths with the presence of pterygium and contracture of neck preventing neck extension. We could insert size 2 LMA without difficulty.

We preserved the spontaneous ventilation during anesthesia by avoiding the use of muscle relaxant. Gamma subunit of AChR plays an important role in the action of muscle relaxants, which is mutated in MPS.[4] The safety of the use of muscle relaxants in these patients is not established thus the use of muscle relaxants was avoided in this case.

Conclusion

There should be a proper preoperative clinical evaluation of pediatric patients with multiple abnormalities to prevent any intraoperative and postoperative complication. The difficult airway should always be anticipated in all cases of MPS, and multiple airway management plans should be available. The patient assessed previously should be reassessed again as airway management becomes more difficult as the age of child increases, so a previously normal airway may become extremely difficult in the future. The anesthesiologist should be aware of challenges associated with this rare syndrome to provide safe anesthesia to these patients.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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