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Escobar Syndrome: A Rare Case and Review of Literature

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Abstract

Escobar syndrome, also known as multiple pterygium syndrome is a rare autosomal recessive disease characterised by presence of web in all flexion joints of the body. Here we present a case of a ten year old female child who presented with multiple contracture in all four limbs and was planned for quadricepsplasty surgery. The child had multiple comorbidities such as thoracic scoliosis, contracture at wrist, ankle and knee joint, bilateral sternocleidomastoid contracture leading to torticollis, thus both bag and mask ventilation and intubation were difficult. Patient was induced with sevoflurane and oxygen and airway was secured with laryngeal mask airway in view of restricted neck extension. Muscle relaxant was not used due doubt of predisposition of such cases to malignant hyperthermia. Bilateral femoral nerve block was given for postoperative analgesia. The surgery was uneventful and the patient was pain free and discharged on 4th postoperative day.

Keywords: *Escobar syndrome; Torticollis; scoliosis; Difficult airway*

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Introduction

Escobar syndrome also called multiple pterigium syndrome is a rare autosomal recessive disorder [1]; was defined as an entity by Escobar., *et al.* in 1978 [2]. Only 60 cases have been reported till now [3] and less than 15 cases of anaesthetic management are there. Questions on management especially whether these patients are predisposed to malignant hyperthermia still remain unanswered and more data is needed to comment on this. Hallmark of the disease is presence of multiple pterigea or contractures across multiple joints (neck, elbow, popliteal area etc). Characteristic faces include ptosis, downward slanting palpebral fissures, hypertelorism, low-set ears and cleft palate. [4] Micrognathia, lingua cochlearis, ankyloglossia may also be present. Orthopaedic deformity in form of arthrogryposis, camptodactyly, syndactyly, arachnodactyly, rocker bottom feet, and vertical talus are commonly seen. [4] Cardiac, urologic and digestive system abnormalities may also be present. Hypotonia, growth retardation, short stature, cryptorchidism, nail anomalies and severe muscular atrophy may also be found. Surgery are commonly required for orthopaedic deformity, genital abnormalities, umbilical or inguinal hernia or adhesion of tonsil to tongue. [5]

Case

A 10 year old female, diagnosed case of escobar syndrome, was scheduled for bilateral quadricepsplasty. She presented with contractures at elbow, phalanges of hands, toes, knee joint and vertical talus (Figure 1).

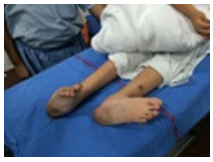

Figure 1

She had deviation of mouth and torticollis with moderate restriction of neck extension due to webbing of neck making airway management difficult.

The child also had dorsolumbar scoliosis (Figure 2) with cobbs angle of 40 degree though there no respiratory compromise. For this she was being managed conservatively with Milwaukee brace. Her ECG showed T wave inversion in V1 to V5 but was asymptomatic and echocardiography was normal. All other investigation were normal.

Standard ASA monitors were attached and induction was done with 8% sevoflurane with 100% oxygen. Airway was secured with laryngeal mask airway. As her knee had never been flexed earlier, the surgeons requested good postoperative analgesia which was given by bilateral femoral nerve block using 0.25% ropivacaine mixed with 0.05 mg/kg dexamethasone. Volume controlled mode was used and anaesthesia maintained with sevoflurane and oxygen nitrous. Recovery was uneventful and the patient remained pain free till discharge on 4th day.

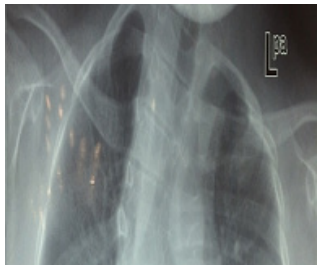

Figure 2

The child was again posted for suturing following quadricepsplasty 3 weeks later. The same anaesthetic technique was used and surgery and postoperative recovery were both uneventful.

Phenotypic Characteristics	Anaesthetic Concerns
Neck contracture and limited neck extension, ankyloglossia, cleft lip and palate	Difficult bag and mask ventilation
Neck contracture, micrognathia, cleft lip and palate, cervical spine fusion	Difficult intubation
Scoliosis and kyphosis	Difficult ventilation and restrictive lung disease, difficult spinal epidural
Limb contracture	Difficult intravenous access

Difficult positioning due to multiple contractures	Difficult peripheral nerve block
Growth retardation	Inability to follow commands upon awakening, difficult extubation
Hypotonia, growth retardation,? prenatal myasthenia gravis	Concern over use of muscle relaxant? malignant hyperthermia

Table 1: *Phenotypic characteristics and anaesthetic concerns arising due to them.*

Paul J. Kuzma., *et al.* [3]	Intubated using fibreoptic after the insertion of LMA and concluded that airway becomes difficult with increasing age.
Ayse Hande Arpaci., *et al.* [6]	Used LMA in a child undergoing frontal sling operation for ptosis and amblyopia.
Ludmyla Kachko., *et al*. [7]	Reported use of general anaesthesia and intubation with the help of intubating stylet (CL grade 3) and epidural for postoperative pain relief.
Shaji Mathew., *et al*. [8]	Reported failed intubation in a 1 yr child and subsequently used proseal LMA followed by railroading ETT using fibreoptic bronchoscope for undergoing bilateral hamstring to quadriceps transfer
Nezih Sertoz., *et al*. [9]	Used combined spinal epidural for surgery and gave low dose spinal (7.5 mg) owing to kyphoscoliosis and chances of rising of block. CSE was given as the airway was difficult
James F . Mayhew., *et al*. [10]	Used GA with Caudal with 2% ropivacaine and 10 lg of preservative free duramorph
Andrea H Jansen., *et al*. [12]	reported the use of the Shikani intubating stylet for the airway management of a neonate with popliteal pterygium syndrome and syngnathia
Caroline Gahm., *et al*. [13]	Discussed the use of Intranasal flexible fiberoptic intubation in a neonate with with extensive interalveolar syngnathia. The patient was sedated with fentanyl and local spray of lignocaine was used.
L. K. Robinson., *et al*. [14]	Reported malignant hypertherima in a 33 week old child with multiple pterigium syndrome after using halothane anaesthesia

Table 2: *Case reports of anaesthetic management of patients with Escobar syndrome.*

Discussion

Escobar syndrome presents multiple challenges to an anaesthesiologist. Main concern of the anaesthesiologist includes difficult airway due to micrognathia, cleft palate, syngnathia, restricted neck movements due to skin folds in neck leading to difficult intubation which was also present in our case. [1,5,6] So laryngeal mask airway was used. Airway management also becomes increasingly difficult with increasing age. Regional anaesthesia may also be difficult due to presence of khyphosis or scoliosis. [7,8] However case reports of successful spinal and epidural are present. [9] Caudal has also been used by Mayhew., *et al*. [9] Recently ultrasound guided spinal epidural has been proposed for difficult cases. Another concern in kyphoscoliosis is restrictive lung disease. Cervical spine fusion may also be present in few cases. [6]

Defect in gamma subunit of acetylcholine receptor [11] has been documented as the cause of escobar syndrome. It has been suggested as a variant of prenatal myasthenia gravis. So muscle relaxant was not used in our case similar to that reported by Arpaci. [6] Intravenous access may be difficult due to presence of contractures. Risk of malignant hyperthermia is still controvercial. [3,10]

Conclusion

Anaesthetic concerns of escobar syndrome thus includes-difficult intubation, difficult intravenous access, difficulty in placing spinal epidural, difficult positioning, concern about use of neuromuscular blockage and a high suspicion for malignant hypothermia. Nerve block thus serves an excellent postoperative analgesia in escobar syndrome without predisposing the patient to any additional risk.

Conflict of interest

There is no financial interest or any other conflict of interest.

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