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Escobar Syndrome: Difficulties in Anesthesia Management

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Authors' contributions

This work was carried out in collaboration between all authors. Authors AK, ÜK, DY and EÖ managed the anesthesia period of patient during the operation. Authors AK, ÜK, DY, EÖ, BK, Kİ and SÖ designed the case report. Authors AK, ÜK, DY and EÖ wrote the first draft of the manuscript. All authors read and approved the final manuscript.

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Case Study

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ABSTRACT

Introduction: Escobar syndrome (multiple pterygium coli) is a rare autosomal recessive disorder which characterized by various malformations. Regarding anaesthesia management, these patients should be considered due to difficult mask ventilation and intubation and risk of malignant hyperthermia by the anesthesiologist.
Case: An 11-year-old girl, with Escobar syndrome, was scheduled for orthopaedic surgery due to femur fracture. In a preoperative physical examination, the patient had revealed multiple joint flexion contractures. Neck extension of the patient was significantly limited due to the flexion contractures, and the mallampati score was 3, but the mouth opening was sufficient. Preoperative echocardiography and electrocardiogram were normal, and there was no abnormality in the complete blood count, coagulation profile and biochemical analysis.

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The patient was taken to the operating theatre without premedication. Following placement of IV line and preoxygenation, anaesthesia was induced with propofol, rocuronium bromide and fentanyl. After repositioning the patient to the 'sniffing' position and applying external tracheal pressure, the trachea was intubated by the conventional laryngoscope with the help of an intubation stylet. Anesthesia was maintained with propofol and remifentanil infusion. The duration of anaesthesia was 210 min and no problems were encountered intraoperatively.

Discussion: Patients with Escobar syndrome are frequent in childhood and are characterized by multiple joint flexion contractures, pterygia of the neck, antecubital and popliteal areas, cleft palate, micrognathia, syndactyly, camptodactyly, decreased mouth openness, vertebral anomalies, and increased risk of malignant hyperthermia.

In the literature, it is emphasized that anaesthesia management of patients with Escobar syndrome has challenging due to difficult intubation, difficult ventilation and the risk of malignant hyperthermia.

Conclusion: In patients with Escobar syndrome, detailed physical examinations, especially cardiac and respiratory system, should be performed before the anaesthesia and necessary precautions such as difficult airway equipment should be taken in the operating theatre.

Keywords: Escobar syndrome; airway management; malignant hyperthermia.

1. INTRODUCTION

Multiple pterygium syndromes (MPS) was first described by Bussiere in 1902 and defined as an entity by Escobar et al. in 1978 [1]. Two different forms of MPS were reported in the literature according to clinical severity, lethal and non-lethal forms. The first form is characterized by prenatal growth deficiency, contractures, pterygia, and dysmorphic facies which is called Lethal MPS. Lethal MPS is thought to be a rare cause of recurrent mid-trimester pregnancy losses. Infants who survive until birth usually die shortly after birth. A primary cause of mortality is likely to be pulmonary hypoplasia [2]. The second form is Escobar type MPS, also known as Escobar syndrome. Escobar syndrome, which is clinically milder than Lethal MPS, is characterized by multiple joint contractures, multiple skin webs (pterygia) across the neck, fingers, forearms, inner thighs, and back of the knee, kyphoscoliosis, camptodactyly with or without syndactyly, growth retardation, craniofacial dysmorphism, and vertebral segmentation anomalies. Also, cleft palate, deafness, short stature and genital anomalies are frequently present as well as widespread musculoskeletal deformities [3].

Although the underlying aetiology in this disorder is unknown, muscle degeneration and disorganization of myofibrils have been shown in some cases by muscle biopsy. MPS has been linked to a mutation of the CHRNG gene which encodes the subunit of acetylcholine receptors (AchR) [4,5,6]. This AchR is essential for signalling between nerve and muscle cells, which is necessary for movement, so its mutation causes fetal akinesia resulting in pterygium (webbing) formation and congenital contractures of joints [7]. The inheritance of MPS is usually autosomal recessive, but autosomal dominant and X-linked forms have been reported in the literature [8].

In this case report, we present the anaesthetic management in Escobar syndrome.

2. CASE

An 11-year-old girl with Escobar syndrome was admitted to orthopaedic surgery due to femur fracture. Multiple joint flexion contractures, pterygia of the extremities and neck, growth retardation, and vertebral deformities (Fig. 1) were observed in the preoperative anaesthetic examination. In the airway evaluation, it was determined that the neck extension was significantly limited due to the flexion contractures and mallampati score was III, but the mouth opening was sufficient. There was no abnormality in the complete blood count, coagulation profile and biochemical analysis. Preoperative echocardiography and electrocardiogram were normal.

The patient was taken to the operation theatre without premedication. She was monitored with five lead electrocardiography, noninvasive blood pressure (NIBP), pulse oximeter (SpO_2), FiO_2 and $EtCO_2$. To prevent the development of intraoperative malignant hyperthermia; dantrolene sodium, sodium bicarbonate and cold IV solutions were available in the operating

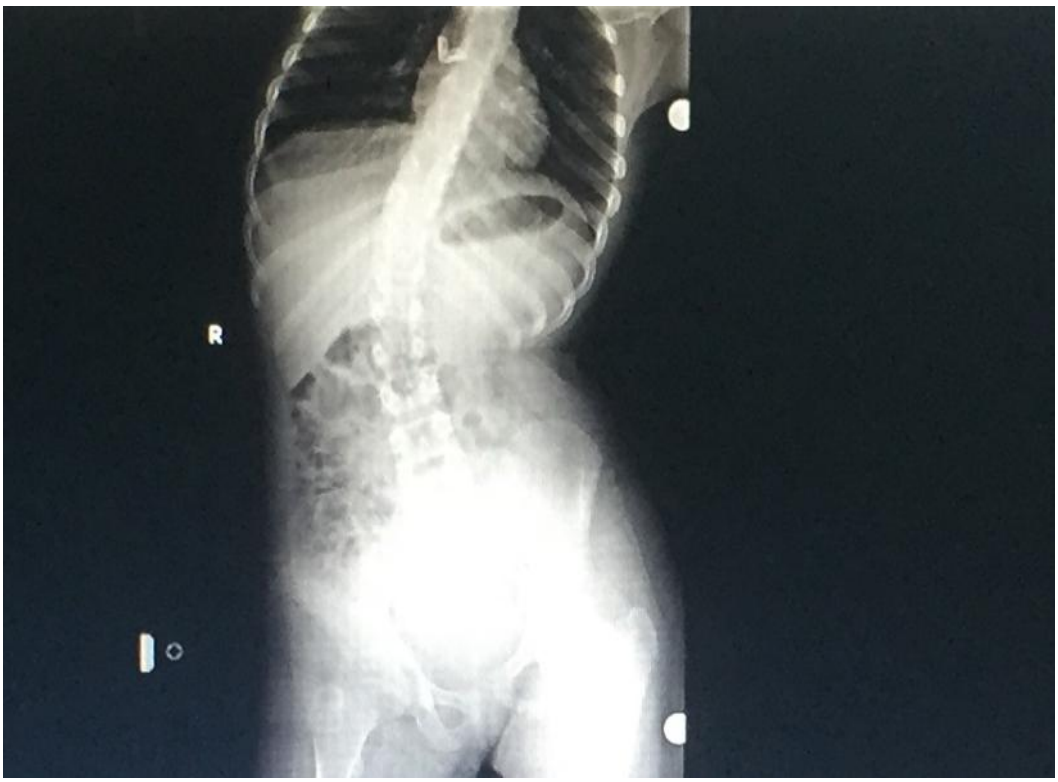

Fig. 1. The radiologic image of our patient which shows vertebral deformity

theatre. We also prepared the equipment for difficult intubation, including flexible bronchoscope, laryngeal masks, different sizes of laryngoscope blades and intubation tubes in the operating theatre. We avoided the use of agents which could trigger MH such as succinylcholine, halothane, enflurane and anticholinergics. Following placement of IV line and preoxygenation, anaesthesia was induced with propofol 60 mg, rocuronium bromide 15 mg and fentanyl 100 µcg. The laryngoscopic view was determined to be Cormack-Lehane Class III using a direct laryngoscope. The first direct laryngoscopy was attempted unsuccessfully. After repositioning the patient to the 'sniffing' position and applying external tracheal pressure, the trachea was intubated with 6.0 mm endotracheal tube with the help of an intubation stylet. Breathing sounds were equal bilaterally and ended tidal CO_2 was detected. During the surgery, body temperature and urine output were monitored. Anesthesia was maintained with propofol and remifentanil infusion. Surgery which was associated with minor blood loss, was completed 210 min. During the surgery, we did not find any increase in temperature and significant hemodynamic changes. Postoperative analgesia was provided by 15 mg/kg paracetamol. At the end of the operation, the patient was transported to intensive care unit as intubated and sedative. All the blood gas and biochemical values of the patient were within the normal limits in the postoperative period. At a postoperative 3rd hour, the trachea was extubated after the patient awakened from anaesthesia. The patient was discharged home on postoperative day 3 in good condition.

3. DISCUSSION

Patients with Escobar syndrome are frequently require an operation because of cleft palate, syndactyly, pes equinovarus, umbilical or an inguinal hernia, scoliosis or other congenital hip anomalies. Escobar syndrome is challenging condition for the anesthesiologist. Patients with Escobar syndrome have increased the risk for potential difficult ventilation and intubation due to the limited mouth opening, cleft palate, syngnathia that in concert with the pterygium colli, limiting the visualization of the pharynx. Difficult regional anaesthesia may occur due to severe scoliosis, kyphosis and other orthopaedic malformations. Possible association with

malignant hyperthermia, difficult IV cannulation and difficult positioning due to multiple contractures may be the other challenging conditions for anesthesiologist [3]. Therefore, detailed preoperative clinical examination, intraoperative and postoperative patient management should be considered very carefully by the anesthesiologist.

It is important to emphasize that airway management of patients with Escobar syndrome becomes more difficult as the patients increase in age and size as a result of an increased deformity of the airway by the pterygia and contractures. Also, rotation of the larynx on its axis also changes with age [3,9]. Therefore, an airway management plan should be established after the conduct of an airway history, and physical examination by the anesthesiologist [9]. Difficult airway devices should be kept ready for these patients before anaesthetic induction. Because, implementation of conventional laryngoscopy, difficulties in glottis visualization and airway management may become difficult in these patients. In the literature, single or multiple successful intubation experiments have been shown in the form of case reports [3,10]. In our patient, risk factors that predicted difficult intubation included diminished neck extension from the pterygium colli, restricted mouth opening, and micrognathia. The first visualization of the larynx revealed Cormack grade III, and the trachea was intubated at the second laryngoscopy, with the help of the 'sniffing position', stylet and external tracheal pressure. If there are difficult airway findings in preoperative clinical examination or intubation is unsuccessful with conventional laryngoscopy in intraoperative period, fiberoptic intubation (FOI), LMA, FOI with LMA can be used for safe and successful intubation in pediatric patients [11,12]. Sethi et al. [3] used LMA without difficulty in their patient with limited mouth opening, short neck with webbing, micrognathia, pectus excavatum, ankyloglossia and pterygia. Kuzma et al. [9] 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. Mathew et al. [13] also used proseal LMA-assisted fiberoptic-guided endotracheal intubation for ventilation. Audenaert et al. [12] used retrograde-assisted fiberoptic intubation which following failed attempts of intubation with direct laryngoscopy and light wand in their pediatric patient with Escobar syndrome. Other alternative techniques such as video laryngoscopy, airtraq optical laryngoscope, and glideslope also successfully used for intubation in pediatric patients with craniofacial anomalies [11,14].

Although it is stated that regional anaesthesia can be applied in appropriate patients, regional anaesthesia is controversial in this group of patients. Kachko et al. [15] reported a case with Escobar syndrome, who underwent bilateral extensive femur osteotomy under combined general and lumbar epidural anaesthesia. Owing to vertebral anomalies such as kyphoscoliosis seen in this syndrome, the rotation of vertebral structures and the depth of epidural space could vary [16,17]. As a consequence, difficulties may arise during regional anaesthesia attempts [15,16], and dural perforation may be observed more frequently [18]. Furthermore, during the regional anaesthesia applications, the sensorial block level may increase in case of using the regular amount of local anaesthetic, compared with normal patients. For this reason, low local anaesthetic volume should be used in these patients. In patients with vertebra anomalies, it is very important to determine vertebral structures under the scope and with ultrasonography [16]. Sertoz et al. [18] applied only regional anaesthesia for their patient with Escobar syndrome who underwent bilateral proximal femur osteotomy and z-plastic. Because their patient's vertebral structure in the lumber region was normal, no difficulties were encountered during the application of the regional anaesthesia. But they applied spinal anaesthesia with low local anaesthetic volume, and combined spinal anaesthesia with epidural anaesthesia to reach appropriate sensory block level when needed.

Malignant hyperthermia (MH) is a substantial threat to the patients undergoing general anaesthesia, requiring the use of non-triggering techniques [10]. Robinson et al. [19] reported a case of Escobar syndrome complicated with MH. However, there is no substantial evidence associating MPS and MH in the literature. In this case presentation, we have considered the risk of MH development and preferred the intravenous anaesthetic agents and monitored the temperature. No change was observed in the perioperative period. But, some authors preferred volatile anaesthetics without complications in the management of these patients with difficult airway findings because of the advantage of

preserving spontaneous ventilation [9,10]. Arpaci et al. [20] used sevoflurane both for induction and maintenance of anaesthesia. Body temperature didn't rise, and hemodynamic stability was maintained under sevoflurane anaesthesia according to their observation.

3. CONCLUSION

Patients with Escobar syndrome must be carefully examined by anesthesiologists during the perioperative period due to the difficulties associated with airway management and MH risk. To determine potential airway difficulties during perioperative period, detailed clinical examinations must be performed; difficult airway equipment must be prepared for intubation and close hemodynamic monitoring must be used during the intraoperative period.

CONSENT

As per international standard or university standard, patient's written consent has been collected and preserved by the authors.

ETHICAL APPROVAL

It is not applicable.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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