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Case Report

Treacher Collins Syndrome: Case Report of a Patient with a Difficult Airway

Anaesthesia Section

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ABSTRACT

Treacher Collin's syndrome (TCS) or mandibulofacial dysostosis is a rare inherited condition which is characterized by bilateral and symmetric abnormalities of the structures within the first and second bronchial arches. The patients with TCS present a serious problem to the anaesthetists in maintaining their airway, as upper airway obstruction and a difficult tracheal intubation due to a severe facial deformity make such a task difficult. Because of retrognathia, the airway management of these patients is often challenging. A 30-year male who was diagnosed with Treacher Collins Syndrome (TCS) was posted for surgery for bilateral temporomandibular joint ankylosis. He presented with dysmorphic facial features, and a difficulty in breathing, sleeping and in lying down since birth. In this case, the most challenging task was induction of anaesthesia and securing the airway.

Key Words: Treacher Collins syndrome, Difficult Airway, Tracheostomy

INTRODUCTION

Treacher Collin's syndrome (TCS) or mandibulofacial dysostosis, is a rare inherited condition which is characterized by bilateral and symmetric abnormalities of the structures within the first and the second bronchial arches. The mechanism of inheritance is autosomal dominant with a variable expressivity. Because of this variability in the expression, some affected individuals exhibit virtually no overt clinical manifestations. However, most of the patients with TCS present with the following classic facial features: down-sloping palpebral fissures, colobomata of the lower eyelid, scanty lower eyelashes, malar hypoplasia, and micro- or retrognathia [1]. The patients with TCS present a serious problem to the anaesthetists in maintaining their airway, as upper airway obstruction and a difficult tracheal intubation due to a severe facial deformity make such a task difficult [2,3]. Because of retrognathia, the airway management of these patients are often challenging [2]. Awake fiberoptic intubation, the well-accepted technique for difficult intubation, can be uncomfortable and stressful for the patient and it requires expertise. Hence, it may not be suitable for the emergency patients and for those who refuse to be intubated awake [4]. Intubating the Laryngeal Mask Airway (ILMA) is a novel method for use in cases of difficult or failed intubation [5].

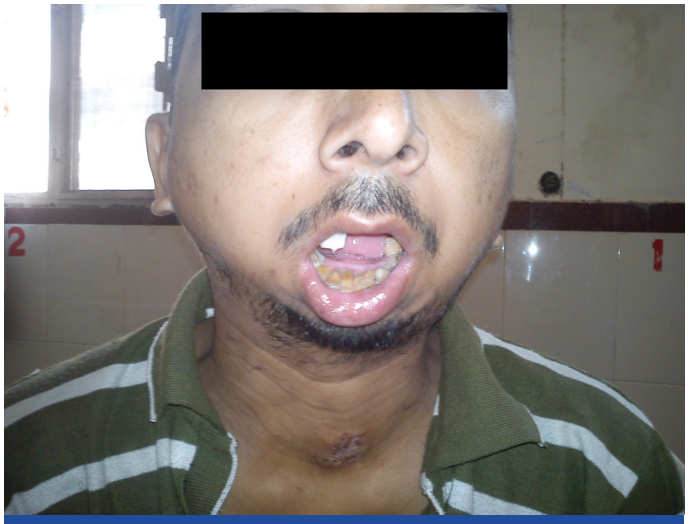
CASE REPORT

A 30-year old male who weighed 50kg, presented with the typical dysmorphic features of micrognathia, zygomatic, mandibular hypoplasia and pectus excavatum. The patient had difficulty in mouth opening, inspiratory stridor, inability to sleep in the supine postion and a history of recurrent pneumonias since birth. The patient was posted for surgical correction of the bilateral bony ankylosis of the temporomandibular joint [Table/Fig-1 & 2].

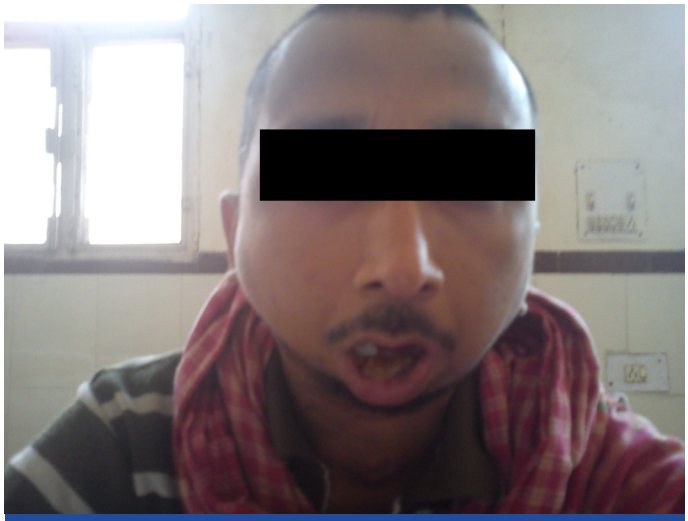
On examination, the patient was found to be afebrile with a heart rate of 82/min and a respiratory rate of 26/min. He was fairly hydrated and he showed inspiratory stridor and chest indrawing with intercostal, subcostal and suprasternal recession. His neck extension was normal. His anaesthetic evaluation showed protruding upper incisors, prominent premaxillas, retrognathia, short thyromental (5cm), sternomental (12cm) and inter incisor distances (1cm), Mallampatti class of IV (6) and Upper Lip Bite Test (ULBT) class III, which predicted a difficult intubation. On chest examination, the pectus excavatum was noted [Table/Fig-3]. X-ray revealed bilateral bony ankylosis of the temporomandibular joint. An elective tracheostomy was planned for the patient.

The patient was prepared with nebulization with Salbutamol and steam inhalation to optimize his respiratory function. The pre operative orders included NPO after midnight, a high anaesthesia risk consent and a tracheostomy consent. The premedications included tab. Ranitidine 150 mg and Alprazolam 0.25 mg which had to be given the night before the surgery and on the pre-operative morning.

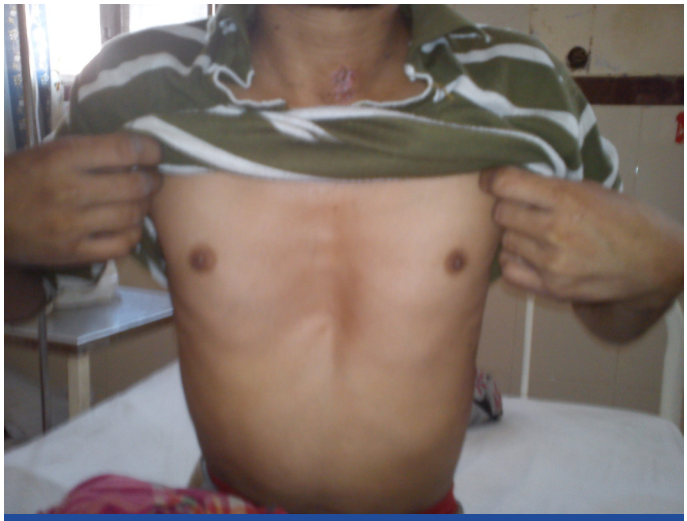
On the day of the surgery, an IV line was secured in the left hand and ringer lactate was started. The pre-medication was done with



[Table/Fig-1]: Patient Photograph 1



[Table/Fig-2]: Patient Photograph 2



[Table/Fig-3]: Patient Photograph 3

glycopyrollate 0.2 mg and tramadol 60 mg and the monitoring which included a SpO_2 probe, ECG leads, a capnograph and a blood pressure cuff were attached after shifting the patient inside the operation theatre. The patient was pre-oxygenated for three minutes with 100% oxygen and anaesthesia was induced with injection propofol 100 mg. A bag and mask ventilation was possible. The blind nasal technique for intubation was tried, to provide ventilation by insufflation as the mouth opening was not possible. The tube was advanced up to the level of the pharyngeal opening, after which a tracheostomy was done by the ENT surgeons to provide the definitive airway. An LMA insertion was not possible due to nil mouth opening, whereas the fiberoptic method and the retrograde technique were not considered as the patient developed respiratory distress in the supine position. A nasal fiberoptic intubation in the sitting position was not considered due to the patient's anxiety.

The Bains coaxial circuit was attached to the tracheostomy tube and anaesthesia was maintained with O_2 and N_2O in a ratio of 40:60 and with halothane 0.5%. The muscle relaxant which was used was vecuronium 0.8 mg/kg .The surgery lasted for 6 hours and the blood loss was minimal. The neuromuscular blockade was reversed with neostigmine 0.05 mg/kg and glycopyrollate 0.01 mg/kg. The patient was shifted to the PACU in the lateral position with a venturimask and he was kept under observation for 24 hours. Oxygen supplementation was done and the vitals were monitored. Adequate post-operative care, pain relief and suctioning of the tracheostomy tube were provided. The tube was removed on the 3rd day and following that, the stoma healed quickly[Table/Fig-3].

The procedures which were followed were in accordance with the ethical standards of the Responsible Committee on Human Experimentation (institutional or regional) and with the Helsinki Declaration of 1975 that was revised in 2000.

DISCUSSION

TCS is a rare genetic disorder which is characterized by craniofacial deformities, the incidence being 1 in 40,000- 70,000 births [6]. It involves the congenital malformations of the first and the second branchial arches, which are inherited as an autosomal dominant trait. These patients have down-sloping palpebral fissures, coloboma of the lower eyelid, scanty lower eyelashes, malar hypoplasia and micro- or retrognathia. Cleft palate is present in up to 35% of the patients and an additional 30%-40% have congenital palatopharyngeal incompetence. Abnormalities of the ear are very common and they vary from minor malformations to severe microtia and hearing loss. The hearing loss may be due to atresia of the auditory canals or ossicular malformation of the middle ear. Despite the multiple development abnormalities, the TCS patients usually have normal intelligence.

The patients with TCS who are posted for various surgeries, pose a problem to the anaesthetists with regards to their airway management. The cause of difficult intubation in such cases is due to relative macroglossia as a consequence of skeletal abnormalities [7,8,9,10]. This reduces the space which is available for the manipulation and insertion of the endotracheal tube. The abnormalities which are associated with TCS may be limited mouth opening, reduced extension of the head on the neck, a hypoplastic mandible and a limited forward movement of the hyoid. Often, multiple mechanisms may be present in an individual case.

Various techniques have been tried for the intubation in TCS viz blind nasal, fiber optic, Bullard's laryngoscope [11], Augistine stylet [12], Shikani optical stylet [13], laryngeal mask airway [14] and laryngeal mask airway with fibreoptic intubation [1]. To improve the success of blind intubation through a laryngeal mask, Brain et al., constructed the Intubating Laryngeal Mask Airway (ILMA) which is being marketed under the name, Fastrach. The new construction allows blind intubation with highly flexible endotracheal tubes to up to 8 mm ID, thus securing the airway around the intubation process and maintaining most of the characteristics of a standard laryngeal mask airway, including contraindications [15].

In this patient, the ASA difficult airway algorithm for an anticipated difficult airway could not be followed, as the patient did not give consent for the trial of awake intubation with the help of local anaesthesia or nerve block, despite having adequate psychological preparations. Also, a retrograde intubation could not be attempted because the supine position was an added hindrance, as it caused respiratory distress. An LMA insertion was not possible because of the nil mouth opening. Hence, the patient's apparent distress and anxiety made tracheostomy with the aid of graded doses of propofol, the suitable choice to secure the airway.

Cricothyrotomy and combitube are emergency procedures for airway maintenance, which are useful adjuncts during a difficult intubation and they appear in the American Society of Anesthesiologists difficult airway algorithm.

CONCLUSION

Craniofacial abnormalities often affect the airway management, making mask ventilation and tracheal intubation difficult or even impossible to achieve, if there are also temporomandibular joint abnormalities. The airway morphology may change as the patients mature or as a result of surgical interventions. The difficulty in the intubation increases as the patients with the Treacher Collins syndrome age and it requires a meticulous assessment of the airway prior to each anaesthetic technique.

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