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Case Report

Anaesthesia Section

Airway Challenges in a Child with Treacher Collins Syndrome having Ventricular Septal Defect, Oculofacial Malformations and Oesophageal Atresia: A Case Report

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ABSTRACT

Treacher Collins Syndrome (TCS), also known as Franceschetti syndrome, is an autosomal dominant condition marked by various developmental anomalies confined to the head and face. It primarily affects craniofacial structures derived from the first and second branchial arches. Patients usually present with down-slanting palpebral fissures, mandibular hypoplasia, lower eyelid colobomas, external ear malformations, and hearing loss. Congenital heart defects and cryptorchidism are two other unusual anomalies. It is typically a bilateral condition. The symptoms and physical features of TCS can vary greatly from person to person. Some individuals may be affected so mildly that their condition goes undiagnosed, whereas others may develop serious, life-threatening complications. Individuals with TCS may require specialised airway management due to a narrow mouth aperture and micrognathia. They are best served by an interprofessional team that can coordinate all aspects of their care. Timely detection and management can improve the quality of life for these patients. The present case report presents the airway challenges in an eight-year-old child with TCS who has congenital malformations and presented with swallowing difficulty, which was later diagnosed as oesophageal atresia.

Keywords: Airway obstruction, Congenital anomalies, Developmental disorders, Oesophageal dilatation

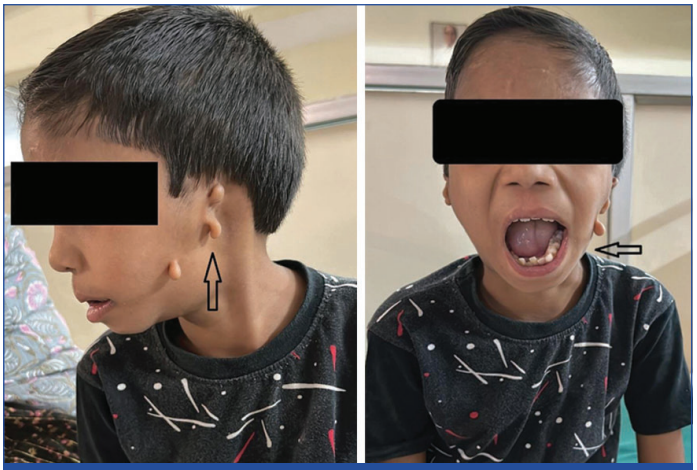
CASE REPORT

An eight-year-old child presented to the casualty with the chief complaint of difficulty swallowing solid food for the last 15 days. The patient also had a history of decreased appetite for the past two months and experienced a generalised tonic-clonic seizure seven months ago. The patient was immediately shifted to the Intensive Care Unit (ICU) where injectable anticonvulsants were administered and then transferred to the ward on the fourth day. The patient had a history of head trauma in a road traffic accident. The birth history of the patient reveals that he was a term baby delivered via lower segment caesarean section, with an eight-day stay in the neonatal ICU. Immunisations are complete and up-to-date. The medical history obtained from the patient's parents indicated that the child had developmental delays, began walking at 2.5 years, started speaking at three years, had oculofacial malformations, and was diagnosed with a Ventricular Septal Defect (VSD) since childhood.

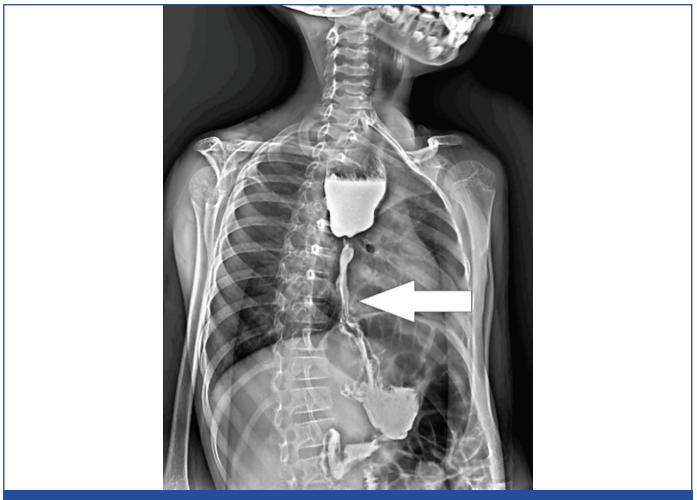
General examination revealed a thin-built, afebrile patient with a pulse rate of 92/min and a respiratory rate of 16/min. The external ear was absent, and the extraoral examination showed malformed ears, microtia [Table/Fig-1], eyes with a downward slant, flattened cheekbones, and a small, steeply angled jaw [Table/Fig-2].

All routine investigations were completed, including a complete blood count, kidney function test, liver function test, and prothrombin time-international normalised ratio. A Computed Tomography (CT) scan of the brain showed asymmetrical pneumatisation of mastoid air cells (left mastoid cells more pneumatised than the right), with the external auditory canal not visualised bilaterally, and a smaller left-side mandibular condyle. An Magnetic Resonance Imaging (MRI) of the brain revealed no abnormalities. Further work-up was conducted to rule out other malformations, with abdominal, pelvic, and renal ultrasonography showing normal results.

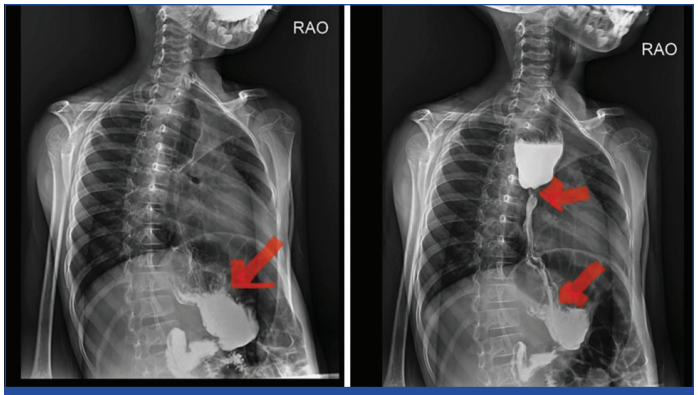
A 2D Echocardiography identified a VSD (closed), with an ejection fraction of 60% and a mildly dilated left ventricle. A barium swallow revealed a proximal oesophageal stricture [Table/Fig-3-5]. Despite



[Table/Fig-1]: Showing malformed ears, microtia. **[Table/Fig-2]:** Small, flattened cheekbones, steeply angled jaw. (Images from left to right)



[Table/Fig-3]: Barium swallow showing oesophageal stricture.



[Table/Fig-4]: Showing distal segment dilatation of the stomach, lower 1/3rd oesophageal narrowing.
[Table/Fig-5]: Upper arrow showing oesophageal dilatation with stricture, lower arrow showing middle 1/3rd and lower 1/3rd oesophageal narrowing. (Images from left to right)

the absence of TCS in the family history, the patient was diagnosed with TCS based on clinical and radiographic findings. The 'sunk-in' appearance and bilateral facial structure involvement, without limb or vertebral abnormalities, differentiate this syndrome from others such as Miller, Nager, and Goldenhar [1]. Since the patient presented with complaints of swallowing difficulty and reduced appetite, following the diagnosis of oesophageal stricture, the patient was scheduled for oesophageal dilatation under Total Intravenous Anaesthesia (TIVA).

A complete preanaesthetic check-up was conducted, and after obtaining paediatric and cardiac clearance, it was determined that a difficult airway was anticipated. During airway assessment, starting from external features, it was noted that the jaw was underdeveloped, making bag and mask ventilation very difficult and preventing a proper seal. The child experienced episodes of apnoea, necessitating the use of a guedel airway and continuous bag and mask ventilation for 20 minutes. Intubation was attempted but unsuccessful, as the vocal cords could not be visualised. Bag and mask ventilation was continued. The patient was found to have a Mallampati score of grade II [2], with an oral opening and thyromental distance of >3 fingerbreadths. Medications were administered based on the patient's body weight. Premedication included Intravenous (i.v.) glycol 0.08, injection Midazolam 1 mg, Ketamine 30 mg, and propofol 40 mg. Upon administration of propofol, the child experienced apnoea, leading to the abandonment of the surgery for three hours. Non operative room intubations were typically more challenging due to the anticipated difficult airway. Given the patient's jaw malformations and difficulties in mask ventilation, head tilt, and jaw lift maneuvers were performed.

After four hours of monitoring, the patient was taken for the procedure again. Endoscopy revealed a tight stricture noted at 22 cm from the incisor teeth. A paediatric endoscope could not be passed across the narrowing into the stomach. Therefore, a Savary Gillard wire was guided across the narrowed segment under fluoroscopic and endoscopic guidance. Graded dilatation was carried out with Savary Gillard dilators ranging from 5 mm to 11 mm, and a similar procedure of oesophageal dilatation was performed after four weeks. Following the lower oesophageal stricture release four weeks later, the upper oesophageal stricture was reduced. The child's discomfort in swallowing solids and liquids decreased, and the child found more comfort with a liquid diet.

DISCUSSION

The TCS, known as Franceschetti-Zwahlen-Klein syndrome, is a craniofacial development disorder with high penetrance. Up to 60% of TCS cases have no family history and are caused by a de novo mutation [3]. TCS is linked to abnormal differentiation of pharyngeal arches during foetal development. Characteristics of TCS include microtia with conductive hearing loss, slanting palpebral fissures with possible coloboma of the lateral part of the lower eyelids, midface hypoplasia, micrognathia, and sporadically cleft palate and choanal atresia or stenosis [4].

The TCS patients may be challenging to clinically diagnose because they exhibit similar features to other disorders such as Miller and Nager syndromes, and the oculoauriculovertebral spectrum (known as Goldenhar syndrome). These disorders are caused by abnormal development of the first and second branchial arches during embryonic development [1].

The TCS is caused by mutations in the TCOF1 and POLR1C genes, which affect facial development before birth [5,6]. TCS is a congenital malformation of the first branchial arch, also known as mandibulofacial dysostosis [6,7]. Congenital heart disease is a significant association in the case of TCS, as shown by the index case. Patients are prone to postoperative upper airway obstruction, pharyngeal oedema, laryngeal oedema, and respiratory distress [8]. Due to upper airway obstruction, relative macroglossia, retrognathia, and difficult intubation in these patients, airway management is challenging. Airway difficulty increases with age, necessitating a re-evaluation of the airway before each anaesthetic [9].

The TCS causes periorbital defects, hypoplasia of zygomatic arches, and deformities of the middle ear. Infants with severe mandibular hypoplasia and airway narrowing may require a tracheostomy tube to breathe [10]. Previous research on other craniofacial syndromes has found that choanal atresia and various forms of cleft palate impair eating and drinking abilities [10]. Therefore, it is reasonable to assume that feeding and swallowing difficulties in TCS are caused by a combination of craniofacial abnormalities [11]. Additionally, feeding difficulties are believed to be the cause of the often reported slim posture in TCS patients [12]. Wong KR et al., discovered malar deformity on Computed Tomography (CT) in TCS patients and found a degree of zygomatic hypoplasia with a decrease in malar volume compared to healthy individuals. Moreover, it was found that decreased malar volume was associated with masseter muscle hypoplasia [13].

A case involving a 13-year-old girl with facial and dental deformities, alongside mild hearing loss, was diagnosed with TCS, showing features of a bird-like appearance of the mandible, hypoplasia, and a high-arched palate, and was treated with supportive care. The deformities in this case were bilateral [14]. Another case involved a 31-year-old individual who presented primarily with a dental anomaly and a history of surgery for a larger mouth and an ear infection, similar to the present case, and was appropriately addressed. These instances contribute to the literature suggesting that TCS patients should be monitored from an early age as they may require multiple surgical corrections in the future for an improved quality of life [15].

A unique case of oesophageal atresia with TCS was reported in a full-term female baby presenting with respiratory distress and micrognathia initially. It was observed that the baby had a hypoplastic midface, absence of lower eyelid lashes, posteriorly displaced tongue, choanal atresia on the left-side, and conductive hearing loss. Treatment to correct the deformities commenced immediately, and the baby responded well and was discharged [16]. In a similar case involving a paediatric patient with TCS exhibiting limited mouth opening, the patient experienced significant airway obstruction during deep inhalation anaesthesia for a dental procedure. Due to the limited mouth opening, an LMA was not used, and the patient was sedated with midazolam and sevoflurane, followed by awake fiberoptic intubation [17]. Mendelian genetic laws state that an affected parent of either sex has a 50% chance of passing on the defect to their offspring. This emphasises the importance of genetic counselling for affected individuals. TCS currently has no known cure, and treatment is tailored to each patient's unique manifestations and requirements. An interprofessional team approach is considered the most beneficial. Correcting various TCS deformities is typically associated with differences in facial

growth patterns and functionality, and many may not necessitate surgical correction [18]. Physicians play a crucial role in recognising this disorder, understanding its manifestations, and providing close follow-up, appropriate therapy, and counselling. Early diagnosis of TCS allows for timely and suitable treatment of the aesthetic and functional deficiencies in these patients [19,20].

CONCLUSION(S)

The TCS presents a significant challenge to anaesthesiologists in terms of airway maintenance due to upper airway obstruction and difficulties in tracheal intubation caused by facial deformities. Airway management in these patients is often complicated by retrognathia. The present case underscores the importance of being prepared for patients with TCS undergoing surgery, as they may have challenging airways, difficult intubation, and may require a trolley and postoperative ICU bed with ventilator support ready. Proper preoperative planning, including thorough examination screening, should be conducted before making a final decision. Since each TCS patient is unique, treatment should be formulated by a multidisciplinary team. Managing TCS is time-consuming and requires a multidisciplinary approach focused on symptom management.

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