Clinical Reports

Anaesthesia for Treacher Collins and Pierre Robin syndromes: a report of three cases

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We present three patients with Treacher Collins or Pierre Robin syndromes who had historical and physical evidence of airway obstruction, difficulty feeding, and sleep disturbances. These preoperative findings correlated with difficult airway management intraoperatively. Based on this experience, we recommend that children with obstructive symptoms have laryngoscopy prior to anaesthetic induction. If the glottic opening is visualized, inhalational induction can proceed. If the glottic structures cannot be visualized, then the anaesthetist must choose between awake oral or nasal intubation, elective tracheostomy, or fiberoptic intubation. In all cases, a tracheostomy tray should be ready and a surgeon experienced in paediatric tracheostomy should be in attendance. After intubation, anaesthesia is best maintained with oxygen and a potent inhalational agent. Extubation should only be done with the patient fully awake and with emergency airway equipment immediately available. Postoperatively, these patients should be transferred to an intermediate care area or intensive care unit where they can be observed closely since delayed complications of airway obstruction are common in this group of patients.

Key words

GENETIC FACTORS: Treacher Collins syndrome, Pierre Robin anomaly, mandibulofacial dysostosis; COMPLICATIONS: cor pulmonale, airway obstruction.

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The Treacher Collins syndrome and Pierre Robin anomaly are congenital malformations of craniofacial development which may present challenging airway management problems throughout the perioperative period. Treacher Collins syndrome is a form of mandibulofacial dysostosis characterized by downward oblique slanting of the palpebral fissures, micrognathia, and cleft palate.[1] Hypoplasia of the maxilla is usually present, and dysplasia of the auditory canals and auditory ossicles result in conductive hearing loss or total deafness.[2,3] Pierre Robin syndrome is classically described as a triad of micrognathia, cleft palate, and glossoptosis (tendency of the tongue to fall downward and backwards).[4,5] Both syndromes can be associated with significant upper airway obstruction even in the unanaesthetized state which can lead to complications such as the sleep apnoea syndrome and cor pulmonale.[6,7] Congenital anomalies of the heart (ventricular septal defect, patent ductus arteriosus, atrial septal defect, and coarctation of the aorta) as well as choanal atresia have also been reported.[2] With the exception of single case reports in the literature,[6,8–11] very little has been written about the anaesthetic management of the patient with Treacher Collins or Pierre Robin syndrome. We describe three such infants and children with significant medical complications of their upper airway obstruction. Anaesthetic choice and airway management techniques are discussed for each individual case and general recommendations are made regarding perioperative care.

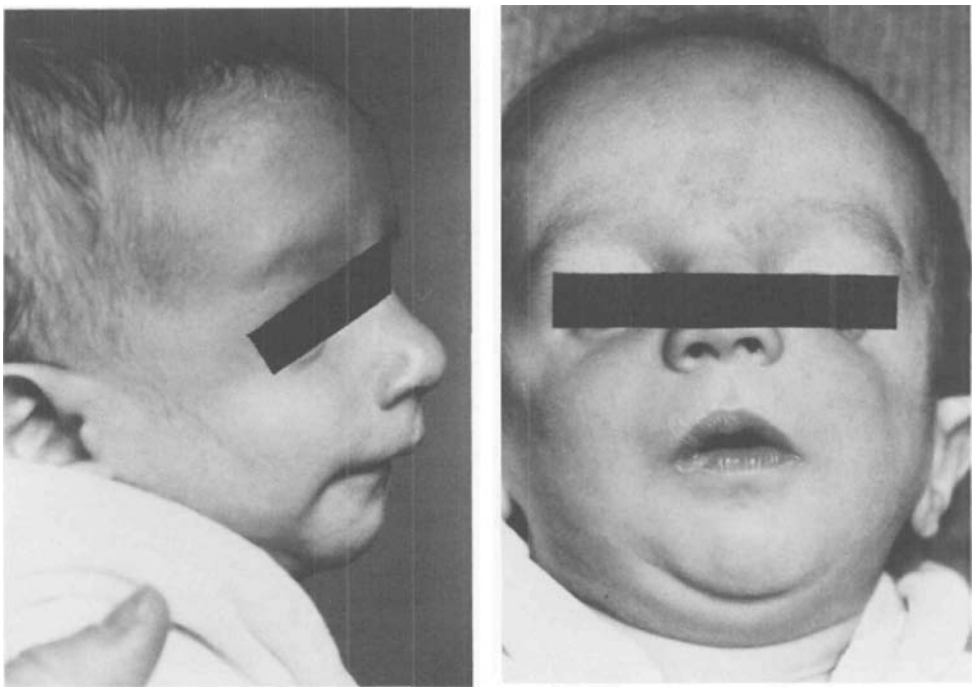

FIGURE 1 Patient with Treacher Collins syndrome (Case I).

Report of cases

Case 1

A seven-week-old male infant with Treacher Collins syndrome, complicated by a hamartoma of the tongue was admitted for removal of the mass and a Beverly Douglas procedure (suturing of the tongue to the lower lip with anterior suspension of the tongue using a suture placed around the mandible and anchored with a button at the base of the tongue).[12] Inspiratory and expiratory stridor with occasional gasping, made worse by supine positioning, had been present continuously since birth. Physical examination revealed an emaciated infant with characteristic facies (Figure 1) in moderate respiratory distress. Weight was 3.96 kg (< third growth percentile for age). The infant had supraclavicular, subcostal, and intercostal retractions that did not improve with positioning, even when prone. Upper airway stridor and rhonchi were present. Examination of the airway revealed a 1×1 cm, hard midline mass on the posterior portion of the tongue, a left unilateral incomplete cleft palate, and some asymmetry of the maxilla. Cardiac exam was normal. The liver margin was 2 cm below the right costal margin. An ECG met criteria for mild right ventricular hypertrophy with an axis of +180.

The infant was not premedicated. Monitors consisting of ECG, precordial stethoscope, and automatic blood pressure device were applied. An intravenous line was inserted. The infant was preoxygenated while being held in the sitting position, leaning forward. Despite cricoid pressure, only the very tip of the epiglottis could be visualized during laryngoscopy and the mass further hindered passage of the endotracheal tube. Oxygen was insufflated during laryngoscopy from a separate source at a rate of $2\ L \cdot min^{-1}$ via a 5 FR feeding tube attached to the side of the laryngoscope blade. The trachea was intubated with a 3.0 mm uncuffed endotracheal tube on the sixth attempt. The infant was anaesthetized with isoflurane in oxygen and spontaneous ventilation was maintained. No muscle relaxants were given.

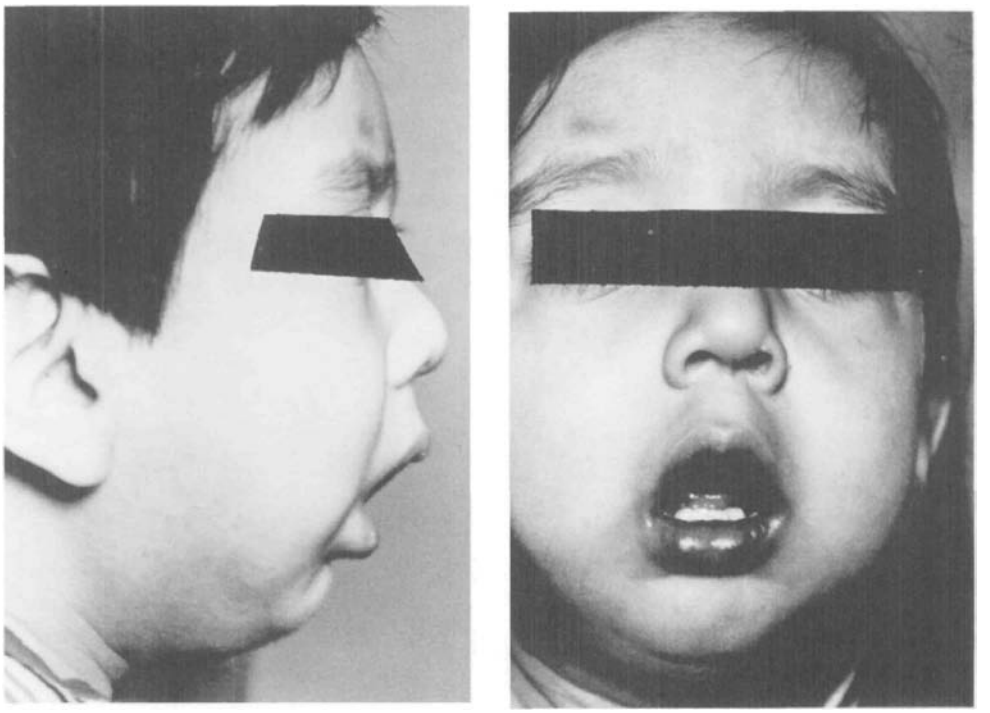

FIGURE 2 Patient with Pierre Robin syndrome (Case II).

The tube was taped and held securely by the anaesthetist. Despite these precautions, during resection of the hamartoma the endotracheal tube was dislodged. The infant made weak respiratory efforts but no breath sounds could be heard. Initially, ventilation with oxygen using a bag and mask was unsuccessful. However, when the surgeon maintained outward traction on the tongue using a retaining suture, ventilation of the lungs improved dramatically. Reintubation orally under direct vision was guided by air bubbles exiting from the larynx. None of the glottic structures could be visualized. The remainder of the case proceeded without difficulty, and the patient was extubated awake in the operating room without complication. The postoperative course was uneventful and the patient's respiratory status has improved significantly.

Case II

A 16-month-old male infant with Pierre Robin anomaly complicated by severe upper airway obstruction and failure to thrive who was admitted for a Beverly Douglas procedure. He had undergone cleft palate repair four months previously at which time the anaesthetist encountered difficulty maintaining an airway during mask induction but had no problems with intubation. Mild to moderate respiratory distress had been present since birth with snoring, stridorous respirations in both the supine or prone positions. He had learned to sleep in an upright position by hanging both arms over the crib rail to hold himself up. Intermittent wheezing and borderline cardiomegaly on chest radiographs suggested pulmonary hypertension and mild cor pulmonale.

The patient was a small anxious-appearing infant with characteristic micrognathia and glossoptosis (Figure 2). Weight was 8.2 kg (< third growth percentile for age). Chest exam revealed an increased A-P diameter, hyperresonant lung fields to percussion, and intercostal retractions at rest. Aus-

cultation of the chest revealed transmitted upper airway rhonchi. The precordium appeared hyperdynamic but no murmurs or gallop rhythm were identified. The liver edge was 3 cm below the right costal margin and the spleen tip was palpable 1 cm below the left costal margin. ECG showed RVH with tall peaked P waves indicative of right atrial enlargment, which was confirmed by M-mode echocardiogram.

The patient was not premedicated. In the operating room, monitors were placed with the infant in the sitting position and an intravenous catheter was inserted under local anaesthesia. After preoxygenation, the patient was placed supine and laryngoscopy performed. The epiglottis and vocal cords were easily visualized; therefore, anaesthesia was induced with halothane in oxygen by face mask with the patient spontaneously breathing in the sitting position. Partial airway obstruction was relieved by insertion of an oral airway. The patient was then placed in the supine position and intubated orally without difficulty using a 4.0 mm endotracheal tube and Miller 1 blade. Anaesthesia was maintained using isoflurane in oxygen via the endotracheal tube, and the patient tolerated the procedure well. At the conclusion of surgery, the infant was extubated when fully awake. At this point, he immediately developed airway obstruction that could not be relieved by positioning, positive pressure, or an oral airway. Although inserting a nasopharyngeal airway fashioned from a 4.0 mm endotracheal tube improved ventilatory status, his continued respiratory distress necessitated reintubation of the trachea. This was accomplished using a blind nasal approach. Although no oedema of the anterior tongue was apparent, there was a haematoma and oedema of the posterior third of the tongue. Therefore, since we anticipated a further increase in airway oedema over the next 24–48 hours and reintubation had been very difficult, elective tracheostomy was performed because of the concern for self-extubation. The tracheostomy tube was removed on the fifth postoperative day and the child has done well since, showing improved weight gain and sleep pattern.

Case III

A six-year-old male with Pierre Robin anomaly and velopharyngeal incompetence admitted electively for a pharyngeal flap procedure. The patient had undergone general anaesthesia five times previously at another hospital and the mother was unaware of any complications with airway management. On physical examination, the child weighed 38 kg (fifth growth percentile for age). He had micrognathia but the tongue did not appear inordinately large. The remainder of his examination was normal. Because the patient had no airway obstruction when sleeping in the supine position, he was given oral premedication with pentobarbitone 50 mg. On arrival in the operating room face mask induction was accomplished using halothane in oxygen without development of airway obstruction. However, intubation was extremely difficult and resulted in the dislodging of a deciduous tooth which was removed with forceps before intubation was finally accomplished. The glottic opening was impossible to visualize even with extreme cricoid pressure, so the endotracheal tube was directed anteriorly, under the epiglottis and passed blindly into the trachea. The patient tolerated the surgical procedure well and was extubated without incident when awake in the operating room.

Discussion

Patients with Treacher Collins or Pierre Robin syndromes may present to the anaesthetist for a variety of procedures from infancy to adulthood. Although the natural history of upper airway obstruction in these patients is one of improvement as they grow,[13] at any stage of their development they may be difficult to ventilte with a bag and mask once anaesthetized and almost impossible to intubate. The pathophysiology of airway obstruction is primarily due to a posteriorly situated tongue which significantly reduces the size of the posterior pharyngeal airway. Each time the infant inhales, the tongue is drawn more posteriorly and downward by the force of negative pressure generated by respiration.[13] This "ball-valve" effect of the tongue produces inspiratory obstruction of the airway which can usually be improved by placing the infant prone, in a head down position, or in a sitting position leaning forward. These manoeuvres encourage slight forward movement of the tongue in response to gravity which improves the obstruction.

Anaesthetic management of these patients should therefore begin with a careful preoperative history with special attention to the cardiorespiratory system and associated congenital abnormalities. Sleep

disturbances, apnoea, snoring respiration and inspiratory stridor may be present. Feeding difficulties such as easy fatigueability, coughing, or diaphoresis with nursing may all indicate upper airway obstruction. After several months of these obstructive symptoms, peripheral oedema, tachypnoea, perioral cyanosis with crying, and hepatomegaly may develop, suggestive of cor pulmonale. Congestive heart failure due to associated congenital defects such as VSD, ASD, or PDA may also complicate the clinical presentation of these infants.

An indirect measure of the patient's ability to compensate for his airway dysfunction is growth. The more severely affected infant with chronic airway obstruction (Case I and II) will present to the physician with height and weight measurements that are well below the average percentiles for age due to increased caloric expenditures to breathe and feeding difficulties which result in poor caloric intake.

After the history is obtained, a careful review of the patient's medical records is extremely helpful since the patient's family may not always be aware of problems encountered during previous anaesthetics (Case III). Physical examination should include a careful evaluation of the child's airway. Nasal flaring, sonorous breathing, chest retractions and an increased A-P diameter of the chest would suggest that significant upper airway obstruction is present. Especially in the small infant, positioning of the patient should be attempted along with other manoeuvres of the head to define the most favourable position for effective air exchange. In most instances, the prone position slightly head down, effectively improves respiration. In addition to airway examination, auscultation of the chest for evidence of pathologic heart murmurs, gallop rhythm, pulmonary rales or wheezing should be done. Peripheral cyanosis and hepatomegaly should also be noted. If cor pulmonale or other congenital heart defects are suspected by history and physical examination, then preoperative ECG, 2-D or M-mode echocardiogram, and arterial blood gases are indicated. Hematocrit $\geq$ 75th percentile for age and CO_2 retention in the absence of a pathologic heart murmur are suggestive of long-standing upper airway obstruction with cor pulmonale.

The patients at greatest risk for anaesthesia are those who present during infancy, or those who develop complications from their upper airway obstruction such as sleep disturbances, failure to thrive, or cor pulmonale. The two infants presented here had mild to moderate respiratory distress in the awake state which could not be relieved by positioning. Sedative drugs may depress awareness in these patients and precipitate cyanosis or frank respiratory arrest on the ward. Therefore, premedicant drugs should be avoided in those patients that have symptoms of significant airway compromise preoperatively. In the older child without obstructive symptoms, judicious use of these medications can provide adequate sedation for placement of intravenous catheters or further airway evaluation in an otherwise anxious, uncooperative child.

Prior to induction of anaesthesia we feel that direct laryngoscopy should be performed in patients with Pierre Robin or Treacher Collins syndrome who have obstructive symptoms in the unanaesthetized state. This can be accomplished awake in small debilitated infants with topical anaesthesia. Cautious IV sedation using narcotics and topical lidocaine is useful in older infants and children. Furthermore, a seen in case 3, a negative history of airway problems during prior anaesthetics does not preclude current problems. This is especially true if operative procedures on the upper airway have been performed in the interim which may affect the diameter and orientation of the nasopharyngeal airway, such as cleft palate repair or pharyngeal flap procedures.

If the glottis can be viewed during laryngoscopy, then an inhalational induction is undertaken. We avoid the use of nitrous oxide because should obstruction or laryngospasm occur, delivery of an FiO_2 of 1.0 affords more time for various manoeuvres to correct the problem before the appearance of cyanosis or bradycardia. We have found that a nasopharyngeal airway formed from an endotracheal tube is very useful for relieving obstruction as the anaesthetic deepens. The breathing circuit is then attached to the endotracheal tube for delivery of anaesthetic gases and oxygen to the patient during attempts at intubation. Another manoeuvre which can improve ventilation is forward and downward traction on the tongue using a suture. These methods are much more helpful than placement of a Guedel airway in the oropharynx for improvement of ventilation in these patients.

If the glottis cannot be easily visualized or severe obstruction is present in the supine position, awake

intubation should be performed either by direct vision orally or by blind nasal approach in the small infant. When awake direct laryngoscopy is used, we either insert a nasopharyngeal airway or attach a 5 FR feeding tube to the side of the laryngoscope blade to continue to deliver oxygen at a flow rate of 1–2 $L \cdot min^{-1}$ during intubation attempts. In the older infant and child, fiberoptic endoscopy is also an option but it must be utilized early during the intubation process to avoid traumatizing the airway which would make visualization through the endoscope impossible. Another option in the older child would be retrograde passage of a catheter via the cricothyroid membrane. In all cases, a tracheostomy tray should be ready and a surgeon skilled in performing paediatric tracheostomy present.

Intraoperatively, maintenance of general anaesthesia with oxygen plus a potent inhalational agent is most attractive because this technique allows spontaneous ventilation to be maintained in the event that the endotracheal tube should become dislodged. Near fatal results have been reported in patients with Treacher Collins syndrome given succinylcholine to facilitate intubation.[8,11] After administration of this drug, the patients became almost impossible to ventilate. Therefore, we avoid the use of muscle relaxants in this group of patients, even after intubation of the trachea has been accomplished.

At the conclusion of surgery, a decision must then be made whether extubation is feasible. This depends largely on the operative site and any trauma to the airway produced during intubation. Unfortunately, in this particular group of patients, an awake vigorous infant or child does not guarantee an adequate airway once the endotracheal tube is removed. This is especially true when the operative procedure involves the posterior pharynx[14] or instruments used for surgical exposure may have caused swelling of the soft tissues in the posterior pharynx or the tongue. The structures most often responsible for obstruction in these patients are located in that area so that very little oedema in the posterior pharyngeal space is required to produce significant respiratory obstruction.

As seen in Case II, external appearance is not a reliable guide to the condition of the posterior pharyngeal airway. Despite no visible airway swelling, extubation of the infant resulted in immediate obstruction. Two other patients reported in the literature were extubated in the OR without incident, but later developed severe airway obstruction in the recovery room.[6,15] One of these patients required percutaneous transtracheal jet ventilation because reintubation could not be accomplished.[15] Therefore, we recommend that if these patients are extubated, they should be transferred from the postanaesthesia recovery room to an intermediate care area or intensive care unit so that they can be observed carefully for late complications of airway obstruction.

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Résumé

Trois patients avec une histoire et évidence physique d'obstruction des voies aériennes, difficultés à se nourrir, et perturbations du sommeil présentant un syndrome de Treacher Collins ou syndrome de Pierre Robin sont présentés. Ces signes pré-opératoires sont correlés avec des difficultés de maintien des voies aériennes per-opératoires. Basé sur cette expérience on recommande que les patients se présentant avec des symptomes obstructifs subissent une laryngoscopie avant l'induction anesthésique. Si l'ouverture de la glotte est visualisée l'induction par agents d'inhalation peut être faite. Si les structures glottiques ne peuvent être viisualisées alors l'anesthésiste doit choisir entre une intubation orale ou nasale réveillée, une trachéostomie élective, ou une intubation par bronchoscope fibroptique. Dans tous les cas un cabarêt de trachéotomie doit être prêt et un chirurgien capable d'accomplir une trachéostomie doit être disponible. Après l'intubation, l'anesthésie est maintenue de préférence avec l'oxygène et un agent d'inhalation puissant. L'extubation doit uniquement être faite une fois le patient réveillé complétement avec un matériel d'urgence pour prendre soin d'un problème des voies aériennes disponibles. En période post-opératoire, ces patients doivent être transférés à des locaux de soins intermédiaires ou de soins intensifs où ils peuvent être observés étroitement car des complications tardives des voies aériennes sont fréquemment observées dans ce groupe de patients.