Tracheal Cartilaginous Sleeve Association With Syndromic Midface Hypoplasia

Brian J. Stater, MD; Karin P. Q. Oomen, PhD; Vikash K. Modi, MD

Tracheal cartilaginous sleeve (TCS) is a rare congenital airway malformation in which distinct tracheal rings are replaced by a continuous cartilaginous segment. Vertically fused C- or O-shaped cartilaginous rings can extend from the subglottis to the carina or bronchus with little to no pars membranacea posteriorly.1 Tracheal cartilaginous sleeve has been associated with various craniosynostosis syndromes.1-12

Infants with craniosynostosis syndromes often have multiple upper airway anomalies, including midface hypoplasia, that may contribute to upper airway obstruction. Since many of these infants are obligate nasal breathers, nasal obstruction will commonly manifest with profound respiratory difficulty. Surgical procedures to correct these upper airway abnormalities can prove difficult, ultimately necessitating tracheostomy.

Focus on the upper airway can detract from investigating laryngotracheal causes of respiratory compromise.1-12 In this particular group of patients, it is important to consider the possibility of TCS prior to tracheostomy. Tracheostomy in a patient with TCS may require modifications in technique to safely secure the airway.

In this analysis we present 2 new cases of TCS and systematically review all documented cases of this condition. We aim to increase awareness of this anomaly and provide recommendations for both intraoperative and postoperative management of patients with tracheal cartilaginous sleeve undergoing tracheostomy.

Methods

This study was reviewed by the director of human research protection programs of the office of research integrity of Weill Cornell Medical College, and it was determined that the content did not meet the definition of human subjects research and therefore did not require institutional review board review.

We reviewed the clinical records of 2 children with TCS treated at the Division of Pediatric Otolaryngology–Head and Neck Surgery of Weill Cornell Medical College.

We also performed a systematic review to identify all documented cases of patients with TCS found in the medical literature. We retrieved publications in PubMed by relevant search terms in title and abstract fields: “tracheal cartilaginous sleeve” and “tracheostomy.” Our search yielded 10 publications in PubMed, of which we present 2 new cases of TCS and perform a systematic literature review of all documented cases of this condition. We aim to increase awareness of this anomaly and provide recommendations for both intraoperative and postoperative management of patients with tracheal cartilaginous sleeve undergoing tracheostomy.

We studied the clinical records of 2 children with tracheal cartilaginous sleeves and short tracheas treated at our institution. One of these patients had Beare-Stevenson syndrome, and the other had Crouzon syndrome. Both patients required tracheostomy for persistent upper airway obstruction, and both required custom-length tracheostomy tubes. Bronchoscopy and needle localization were beneficial in performing tracheostomy on these patients. All documented cases of tracheal cartilaginous sleeve in the literature were found to be associated with midface hypoplasia secondary to syndromic craniosynostosis. Seventy-five percent of cases required tracheostomy.

Tracheal cartilaginous sleeve can be associated with Beare-Stevenson syndrome. In infants with midface hypoplasia associated with a craniosynostosis syndrome undergoing tracheostomy, the surgeon must be prepared to encounter and manage tracheal cartilaginous sleeve intraoperatively. Bronchoscopy and needle localization can be beneficial when performing tracheostomy in these patients. Customized, shorter-length tracheostomy tubes should be considered for these patients.

In this study, we report 2 new cases of tracheal cartilaginous sleeve and perform a systematic literature review of all documented cases of this condition. We aim to increase awareness of this anomaly and provide recommendations for both intraoperative and postoperative management of patients with tracheal cartilaginous sleeve undergoing tracheostomy.

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Research Case Report/Case Series

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Infants with craniosynostosis syndromes often have multiple upper airway anomalies, including midface hypoplasia, that may contribute to upper airway obstruction.2 Since many of these infants are obligate nasal breathers, nasal obstruction will commonly manifest with profound respiratory difficulty. Surgical procedures to correct these upper airway abnormalities can prove difficult, ultimately necessitating tracheostomy.

Focus on the upper airway can detract from investigating laryngotracheal causes of respiratory compromise.1-12 In this particular group of patients, it is important to consider the possibility of TCS prior to tracheostomy. Tracheostomy in a patient with TCS may require modifications in technique to safely secure the airway.

In this analysis we present 2 new cases of TCS and systematically review all reported cases of TCS found in the medical literature. In addition, we propose an intraoperative technique that aids in performing tracheostomy on these patients.

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which 7 were included in this study. Cadaveric studies were excluded, as were cases of postmortem diagnosis. Data on clinical diagnosis, associated syndromes, other airway anomalies, need for tracheostomy, surgical technique and complications were collected for all documented cases.

Report of Cases

Case 1

A 3600-g term girl was born with clinical stigmata of Crouzon syndrome, including craniosynostosis and midface hypoplasia. Fiberoptic nasal endoscopy could not be performed secondary to nasal cavity stenosis. She developed respiratory distress several weeks after birth and required mechanical ventilation. A computed tomographic (CT) scan revealed complete nasal cavity stenosis, a thickened bony nasal septum, but patent choanae. Oral flexible fiberoptic laryngoscopy showed mild glossoptosis and no laryngeal anomalies.

The patient had continuous respiratory difficulties during the first year of life, requiring repeated intubations and mechanical ventilation. A computed tomographic (CT) scan revealed complete nasal cavity stenosis, a thickened bony nasal septum, but patent choanae. Oral flexible fiberoptic laryngoscopy showed mild glossoptosis and no laryngeal anomalies.

The patient had continuous respiratory difficulties during the first year of life, requiring repeated intubations and mechanical ventilation in the setting of viral upper respiratory tract infections. Direct laryngoscopy and bronchoscopy at age 3 months revealed a short trachea with poorly defined tracheal rings, a normal pars membranacea, and a rotated carina.

Tracheostomy was performed at age 10 months for upper airway obstruction that had triggered recurrent hospital admissions. During initial admission at an outside hospital, a noncustomized, age-appropriate tracheostomy tube was inserted. This tracheostomy tube was too long and it resulted in right main stem bronchus ventilation and further respiratory distress. Bronchoscopy at our institution revealed 30% obstructing granulation tissue at the carina. The granulation tissue was treated with ciprofloxacin-dexamethasone drops and resolved.

Case 2

A 2200-g preterm boy was born with multiple features of craniosynostosis including midface hypoplasia, ocular proptosis, hypertelorism, preauricular pits, and palmar cutis gyrata. He was intubated shortly after birth for respiratory distress. A CT scan showed complete nasal stenosis and choanal atresia. Genetic microarray testing revealed Beare-Stevenson cutis gyrata syndrome (BSS) with a confirmed mutation in \( FGFR2 \).

Tracheostomy was performed at age 3 weeks. Direct laryngoscopy and bronchoscopy revealed a short trachea, a nor-

Figure 1. Flexible Tracheoscopic View of Patient 1 Through Endotracheal Tube

Localization of tracheostomy using a needle. Arrowhead indicates pars membranacea.

Figure 2. Direct Bronchoscopic View (Proximal Trachea) of Patient 2

Short trachea and lack of tracheal rings are apparent.
mal pars membranacea, and no discernable tracheal rings (Figure 2 and Figure 3). The exposed airway from the thyroid notch to the trachea appeared to be one continuous rigid cartilaginous sheet with no discernable cricoid or cricothyroid membrane. A needle was placed externally through the airway and confirmed to be correctly positioned distal to vocal folds via a direct bronchoscopy. The tracheostomy was then completed without complication, although it was difficult to incise the trachea because a No. 11 blade scalpel would not sufficiently penetrate.

Review of the Literature
We were able to identify 34 cases of documented TCS in the existing literature. Combined with our 2 patients, a total of 36 patients with TCS have been described, of whom 75% (n = 27) required a tracheostomy (Table).

Each case of TCS occurred in the setting of midface hypoplasia associated with a craniosynostosis syndrome. The most commonly associated syndrome was Crouzon, followed by Pfeiffer and Apert syndromes. Twelve of 13 patients with Crouzon syndrome and TCS required tracheostomy for definitive airway management. Choanal atresia, nasal stenosis, and abnormally short tracheas were noted as coexisting anatomical abnormalities in this subset of patients. Ten of 11 patients with Pfeiffer syndrome required tracheostomy, whereas only 2 of 7 patients with Apert syndrome underwent tracheostomy. One of our patients was the first reported case of TCS associated with BSS.

Discussion
In the medical literature, all reported cases of TCS have been associated with midface hypoplasia secondary to a craniosynostosis syndrome.\(^1\)\(^{-13}\) Our 2 cases were consistent with this trend.

Beare-Stevenson cutis gyrata syndrome is an extremely rare, autosomal dominant condition that typically presents with skin furrows of corrugated appearance (cutis gyrata), acanthosis nigricans, midface hypoplasia, craniosynostosis, anogenital anomalies, prominent umbilical stump, and skin tags.\(^{14}\) Hall et al\(^{14}\) reported that choanal atresia was a common feature in BSS, appearing in 5 of 6 patients in their case series. Our patient with BSS also presented with choanal atresia. However, TCS has not been reported in patients with BSS.

The rigidity of the trachea in patients with TCS is believed to alter the mechanical properties of the airway and airflow dynamics, reducing the efficacy of natural airway protective mechanisms and airway clearing.\(^4\) Davis et al\(^{4}\) proposed that the TCS lacks the distensibility of the normal trachea, which ordinarily includes the interring fibrous tissue between cartilaginous rings as well as a pars membranacea. Scheid et al\(^{4}\) also propose that the lack of a pars membranacea may detract from the necessary elasticity needed to provide distention. Furthermore, Hockstein et al\(^{7}\) questioned if the airway of a patient with TCS can adequately grow to meet the oxygen needs of a developing child.

Multiple authors have proposed that tracheostomy can confer significant benefit in this subset of patients. Lertsburapa et al\(^{1}\) demonstrated in a meta-analysis that tracheostomy placement in patients with TCS can decrease morbidity and increase survival. However, the rigidity of the trachea and...
lack of normal anatomical landmarks can make tracheostomy challenging in these patients.

In our 2 patients, TCS diagnosis was made on direct visualization of the outer laryngotracheal skeleton while performing tracheostomy. Both patients underwent bronchoscopy prior to tracheostomy, but TCS was not obvious. This concurs with the findings of Lin et al,3 who described 3 cases of TCS, 2 of which were diagnosed postmortem. In the 2 cases that underwent bronchoscopy, TCS was not recognized endoscopically.

However, TCS is potentially an identifiable disorder, and several authors have described its subtle bronchoscopic appearance.1,2,4,7 Inglis et al4 describe anterior vertically fused tracheal rings in 4 patients with craniosynostosis syndromes. In 3 of 4 patients, initial bronchoscopy revealed a smooth cartilaginous anterior trachea without ring segmentation characterized by the absence of mucosal indentations. A normal pars membranacea and a dysmorphic carina were also described. This is consistent with our cases in which bronchoscopy revealed a normal pars membranacea and the absence of tracheal ring segmentation.

To our knowledge, the importance of performing tracheostomy under direct bronchoscopic vision has not been described in the literature. Intraoperative bronchoscopy can serve as an important aid in determining the proper location for tracheostomy in the absence of clear anatomical landmarks, especially important given the frequently shorter-length tracheas found in these patients.

The rigidity of the TCS can make insertion of the tracheostomy tube challenging intraoperatorily; Alli et al2 recommend performing a window tracheostomy, rather than the traditional slit incision, to facilitate tracheostomy tube placement in patients with TCS.

Both patients in our series were noted to have shorter-than-average tracheas and required customized tracheostomy tubes with shorter distal lengths. Hockstein et al7 described similar findings, noting a short trachea in 3 of 5 patients with Pfeiffer syndrome and TCS. Alli et al2 described a patient with Pfeiffer syndrome and TCS undergoing tracheostomy; they did not report the tracheal length in this patient, but they described use of a tracheostomy tube with adjustable distal length. Mixter et al8 described a case of progressive respiratory obstruction due to posterior tracheal wall injury from a tracheostomy tube in a patient for whom TCS was ultimately diagnosed postmortem. Progressive respiratory obstruction may have also occurred due to granulation tissue formation due to a tracheostomy tube of excessive length and inappropriate angle.

One of our patients experienced obstructing granulation tissue distal to the tracheostomy tube tip, just cephalad to the carina, even in the presence of a tracheostomy tube that was adjusted to the patient’s shorter tracheal length. This tendency to develop airway granulation tissue in patients with TCS has been described by several authors.1,2,7,8 Aware of this tendency, several authors advocate increased frequency of elective bronchoscopies in their patients.1,7

From our experience with TCS, we recommend consideration of a customized tracheostomy tube with a shorter distal length for both the short- and long-term postoperative period. Of note, one of our patients experienced a critical complication from receiving right lung ventilation due to the inadvertent insertion of a noncustomized, age-appropriate tracheostomy tube into the right main stem bronchus.

Accidental decannulation in the postoperative period is one of the most feared complications of pediatric tracheostomy. In patients with TCS, who must receive a shorter tracheostomy tube, accidental decannulation is more likely. Reinsertion of the tracheostomy can prove difficult and may result in greater mortality and loss of the airway owing to the rigidity of the trachea and lack of normal laryngotracheal landmarks.

One of our patients experienced an accidental decannulation in the first week after tracheostomy, and rigid bronchoscopic visualization was required for reinsertion of the tracheostomy tube. At our institution, patients with recent tracheostomy are kept in the intensive care unit until the first tracheotomy tube change is performed to verify that the tract is well established and the airway is secure.

Conclusions

Tracheal cartilaginous sleeve can be associated with BSS. In patients with midface hypoplasia associated with a craniosynostosis syndrome undergoing tracheostomy, the surgeon must be prepared to encounter and manage TCS intraoperatorively. Bronchoscopy and needle localization can be beneficial when performing tracheostomy in patients with TCS. Patients undergoing tracheostomy should be considered for customized, shorter-than-average tracheostomy tubes and managed conservatively in the postoperative setting.