# Tracheal stenosis and congenital heart disease in patients with Down syndrome: diagnostic approach and surgical options[☆]

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## Abstract

Upper airway obstruction is a prevalent feature in patients with Down syndrome. However, these patients may be completely asymptomatic in the early months of life. The recognition of a problem within the airway quite often occurs when these children present for cardiac surgery, diagnostic radiography with sedation, or during the induction and intubation for anesthesia. Tracheal stenosis is rare in the general population, but is seen somewhat more frequently in patients with Down syndrome. The incidence of tracheal stenosis in children with congenital heart disease, which is seen in 40% of patients with Down syndrome, has been reported to be 1.2%. Patients with Down syndrome also tend to have other upper airway obstructive pathology such as nasopharyngeal, oropharyngeal, and subglottic compromise. These entities, combined with the high incidence of cardiac disease, put these children at risk for acute and chronic cardiopulmonary compromise. We present two patients with Down syndrome and congenital heart disease who were found to have significant tracheal stenosis at the time of their cardiac surgery. The perioperative management of their airway defects including diagnostic evaluation and treatment modalities are discussed. © 2000 Elsevier Science Ireland Ltd. All rights reserved.

*Keywords:* Down syndrome; Tracheal stenosis; Tracheoplasty; Congenital heart disease

## 1. Introduction

Upper airway obstruction is a prevalent feature in patients with Down syndrome, and may be secondary to oropharyngeal hypotonia, adenotonsillar hypertrophy, macroglossia, micrognathia, reduced nasal airway, short broad neck, subglottic stenosis, or tracheal stenosis [11,16]. Associated cardiopulmonary disease in these children may compound the effects of upper airway pathology. Often, these patients may have unrecognized airway lesions on presentation for evaluation and management of cardiac pathology.

Down syndrome is seen in 1/1000–1/700 live births, with increased incidence associated with advanced maternal age. The syndromic features are secondary to trisomy of chromosome 21 in most cases. In 4% of the cases, there is a translocation pattern, and 1% of the patients have mosaic trisomy 21 chromosomal patterns [13]. Common characteristics of patients with Down syndrome include craniofacial anatomy that may contribute to upper airway obstructive patterns. They may also have concomitant tracheobronchial and cardiac abnormalities.

The incidence of tracheal stenosis in children with congenital heart disease has been reported to be 1.2% [19]. It is also relatively common in patients with Down syndrome. Tracheal stenosis in these patients tends to be of the segmental type and is secondary to absent pars membranacea of the trachea, or complete tracheal rings [1].

We present two patients with Down syndrome and congenital heart disease who first demonstrated airway symptomatology at the time of cardiac surgery. Diagnostic laryngoscopy and rigid tracheobronchoscopy should be an integral component of the diagnostic evaluation. Radiologic evaluation, preferably magnetic resonance imaging (MRI), will rule out possibility of extrinsic tracheal compression secondary to vascular anomalies, or distal tracheobronchial pathology. Options for airway reconstruction include endoluminal tracheal stent placement, tracheal expansion utilizing autologous cartilage, slide tracheoplasty, and end-to-end anastomosis.

## 2. Case 1

A 2-week old female with Down syndrome presented for ligation of a patent ductus arteriosus (PDA). At the time of surgical repair, she was noted to have significant ventilatory difficulty requiring bronchoscopic airway evaluation. Urgent tracheobronchoscopy revealed a 2-cm length region of circumferential tracheal stenosis (approximately 3 mm in diameter), consistent with complete tracheal rings to the level of 1 cm above the carina. She was successfully extubated, but suffered intermittent respiratory distress over the following months. At age 11 months, she underwent concomitant repair of her ventriculoseptal defect (VSD) and her tracheal stenosis. Tracheal repair involved pericardial and goretex patch graft to an anteriorly incised site of tracheal stenosis. Despite significant improvement in the tracheal airway diameter, the patient was unable to be extubated secondary to prolapse of goretex and pericardium into the tracheal lumen. She then underwent placement of an expandable metallic Palmaz (R) (Johnson & Johnson Interventional Systems Co., Warren, NJ) wire stent under bronchoscopic and fluoroscopic guidance. The presence of pericardium and goretex allowed for the utilization of an expandible stent, given that this area of obstruction was no longer a fixed cartilaginous stenosis. Postoperative management has included interval bronchoscopies, balloon dilatations of the stent, and removal of granulation tissue from the wire mesh interspaces. The stent has been dilated to its maximal width of 9 mm (Fig. 1). Her airway diameter has remained stable at a 3.5-year follow-up period.

## 3. Case 2

A 5-month-old female with Down syndrome and no prior airway symptomatology underwent VSD closure, PDA ligation, and closure of a patent foramen ovale (PFO). She was recognized to be a difficult intubation, requiring a small (2.5-mm inner diameter) endotracheal tube and the inability to advance the tube to the appropriate length in the trachea. She developed ventila-

tory difficulty during cardiac surgery. Urgent bronchoscopy demonstrated a 3-cm length mid-tracheal pinpoint stenosis (approximately 2-mm diameter) with complete tracheal rings, 1.5 cm proximal to the carina (Fig. 2). She was returned to the operating room for local tracheal resection with utilization of anterior tracheal cartilage as an onlay patch graft, as described by Backer and Holinger [2]. A longitudinal incision was made from just proximal to the carina, through the
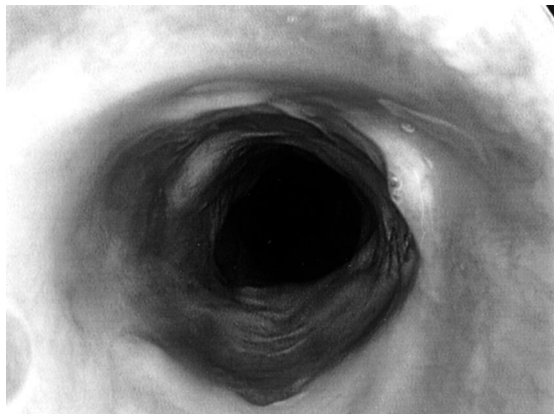


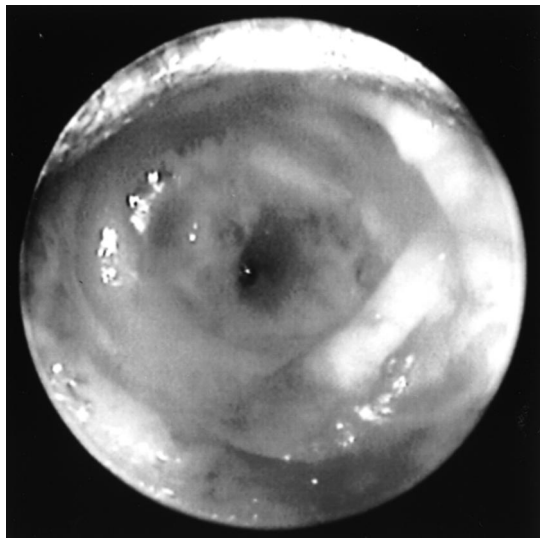Fig. 1. Trachea of Case 1 patient with dilated, mucosalized stent.



Fig. 2. Trachea of Case 2 patient on initial bronchoscopy, demonstrating pinpoint region of tracheal stenosis.
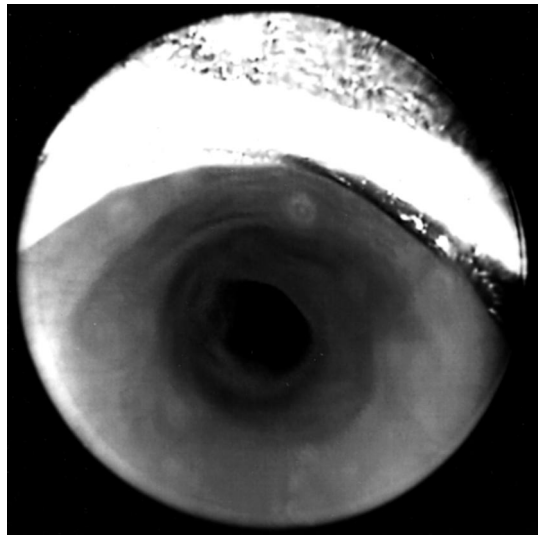


Fig. 3. Trachea of Case 3 patient 2 weeks following anterior cartilage graft, demonstrating enlarged tracheal airway.

stenosis, into normal trachea. The area of tracheal stenosis was resected en bloc. The posterior wall of the tracheal segments was anastomosed end-to-end. The anterior tracheal segment was left open, and the excised tracheal cartilage was utilized as an anterior onlay patch graft. She was extubated under bronchoscopic guidance successfully on postoperative day 15. Bronchoscopy demonstrated significant improvement in airway diameter (Fig. 3). She had persistent intermittent stridor at a 6-month follow-up period, necessitating revision tracheoplasty with segmental tracheal resection and end-to-end anastomosis (without onlay patch graft). A tracheotomy tube was placed to alleviate progressive upper airway obstruction secondary to oropharyngeal hypotonia. She remains stable with a stable tracheal airway at a 2-year follow-up period.

## 4. Discussion

Multiple congenital anomalies are closely linked to Down syndrome. Cardiac malformations are found in 40% of these patients, a large number of whom will require cardiac surgery. Laryngeal and/or tracheal pathology as the source of airway

obstruction is of particular concern, as it will inevitably be exacerbated by intubation of the airway. There is also an increased incidence of subglottic stenosis (6%) in these patients as compared with the general population [11,13]. There are two possible explanations for this. One is that the subglottic stenosis is indeed congenital and represents a component of these patients' generally smaller laryngotracheal airway [1]. The other possible explanation reflects the fact that a large percentage of these patients require one or multiple intubations early in life for cardiac surgery, upper airway obstruction, or diagnostic procedures, leading to iatrogenic subglottic stenosis [11,13].

Endotracheal intubations may lead to chronic inflammation and scarring of the subglottic airway, and patients with Down syndrome should be intubated with an endotracheal tube 0.5–1.0 mm diameter smaller than the standard age-appropriate endotracheal tube size. There are the likely components of both congenital and iatrogenic stenosis contributing to subglottic pathology in these patients [13].

Tracheal stenosis in patients with Down syndrome is unlikely to be of iatrogenic origin. There are three types of tracheal stenosis, as described by Cantrell and Guild in 1964. These include generalized tracheal hypoplasia, which is the most severe form and has the highest associated mortality, 'funnel-like' stenosis, characterized by progressive proximal to distal tracheal stenosis, and 'hourglass', or segmental, stenosis, which can be an isolated segment of mid- to distal stenosis. [3,12] The 'hourglass', or segmental, form is the most common of the three types of tracheal stenosis associated with Down syndrome [18].

Diagnostic evaluation for tracheal pathology in children with Down syndrome should be undertaken in patients presenting with a history of stridor, cyanosis, wheezing, or recurrent pneumonias. Associated feeding difficulties may correspond to a vascular anomaly encroaching on both the trachea and the esophagus. In children who are intubated, trachal stenosis should be considered if there is a history of intubation difficulty or a requirement for high pressure ventilatory support while on a ventilator [15].

The diagnostic evaluation should include radiologic as well as endoscopic modalities. Contrast tracheobronchograms may be performed utilizing isotonic contrast in the trachea to visualize the contour of the tracheobronchial tree. Barium esophagrams will demonstrate the presence or absence of extrinsic esophageal compression. These two modalities are utilized less frequently, with the widespread availability of computerized tomography and magnetic resonance imaging to provide clear images of both aerodigestive and vascular anatomy of the neck and chest [15]. Magnetic resonance imaging is now the imaging modality of choice in the evaluation of tracheal stenosis. Multiplanar images can be visualized, and differentiation between intrinsic tracheal pathology and extrinsic tracheal compression can be assessed. This also allows for visualization of the distal tracheobronchial tree [15]. Accurate assessment of the laryngotracheal airway necessitates airway endoscopy. Rigid tracheobronchoscopy allows for thorough evaluation of laryngeal, tracheal, and proximal bronchial anatomy, while concomitantly enabling dynamic assessment of these areas [15]. Direct endoluminal visualization allows for differentiation between stenotic segments and segments of complete tracheal rings with absent pars membranacea. It also enables visualization of intraluminal versus extraluminal tracheal compression.

There have been multiple interventions performed in attempt to improve the tracheal airway in patients with tracheal stenosis. In patients who are not ventilator dependent, and can tolerate prolonged periods of time without hospitalization for airway support, close observation with aggressive early intervention with medical therapy for upper respiratory tract infections may alleviate the necessity for surgical intervention. However, in children with severe, prolonged airway symptomatology, surgical intervention is warranted. For short segment stenoses, tracheal resections with primary anastomoses have been successful [8]. For longer segments of stenosis, anterior and/or posterior tracheal wall division with placement of autologous tissue grafts have been effective. Grafted tissue may be costal cartilage, pericardium [12], or resected tracheal cartilage utilized

as a patch graft [6]. Slide tracheoplasty has also been useful for long segment tracheal stenoses [5].

Endoscopic endoluminal tracheal expansion techniques are being advanced for both tracheomalacia and tracheal stenosis. These include tracheobronchial angiocatheter balloon dilatation under fluoroscopic guidance [10,14,17] and placement of endoluminal tracheobronchial stents [4,6,7,9,14]. Both have demonstrated variable short- and long-term efficacy. Complications secondary to balloon dilatation include post-dilatation re-stenosis and granulation tissue. Complications secondary to endoluminal stent placements have included persistence of granulation tissue, stent migration, tracheal perforation, tracheal hemorrhage, stent fragmentation [9] and inability to remove the stent secondary to tracheal mucosal overgrowth around the wire mesh of the stent [4].

## 5. Conclusions

Tracheal stenosis is a rare entity in the general population. It is seen somewhat more frequently in patients with Down syndrome. These patients also tend to have other upper airway obstructive pathology such as nasopharyngeal, oropharyngeal, and subglottic compromise. These entities, combined with high incidence of cardiac disease, put these children at high risk for acute and chronic cardiopulmonary complications. Patients with Down syndrome and signs and symptoms of upper airway obstruction should be evaluated closely to identify the level or levels of obstruction. Direct laryngoscopy and rigid tracheobronchoscopy should be an integral component of the diagnostic evaluation. Radiologic evaluation, preferably by MRI, will rule out the possibility of tracheal compression secondary to vascular anomalies or distal tracheobronchial disease.

Options for airway reconstruction include endoluminal tracheal stent placement and tracheal expansion utilizing autologous cartilage, slide tracheoplasty, or pericardium. For short segment stenoses, primary resection with end-to-end anastomosis has been successful.

We report our results in one child utilizing endoluminal tracheal wire stent placement. The long-term concern in this patient is the likelihood that this stent will not be able to be removed endoscopically, given that it is completely incorporated in the tracheal mucosa and pericardial/goretex graft. Her tracheal airway is currently 9 mm in diameter, which is the maximal width of the stent. She will be followed with interval bronchscopic evaluations to determine the long-term outcome of the stent's utility. The second child underwent primary resection of the stenotic segment, first with tracheal cartilage onlay graft, and later with end-to-end anastomosis. One would presume that her expanded tracheal diameter would continue to grow with her growth.

In short, we present the diagnostic evaluation and management of two cases of children with tracheal stenosis who presented during cardiac surgery. The otolaryngologist, anesthesiologist, and cardiothoracic surgeon must be aware of the potential for the presence of laryngotracheal pathology in the management of cardiac disease in patients with Down syndrome.

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