Tracheal agenesis: a case report, including management and review of new treatment strategies

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Abstract

Background: Tracheal agenesis is a rare and fatal malformation. It can be associated with certain syndromes and should be suspected in cases of polyhydramnios in neonates under 2500 grams with respiratory distress. Early recognition is critical to allow for appropriate oxygenation initially through esophageal intubation.

Methods: Case report and review of the current literature.

Results: We report on an unusual case of tracheal agenesis and management of a neonate with an imperforate anus, left polydactyly with an extra toe, and APGAR scores of 3 and 4 at 1 and 5 minutes respectively. Also, mother was found to have polyhydramnios on prenatal ultrasonography.

Conclusions: Tracheal agenesis is a rare developmental phenomenon that is classically incompatible with life. The upper airway is uniquely designed to have rigidity that prevents collapse with respiration and yet is also flexible and pliable to allow for movement with deglutition, coughing, and neck movements. This along with the improvement of its growth capacity and unique ciliated epithelium for mucous discharge makes us think that this is because the tracheal agenesis tube is more rigid and allows for higher pressure ventilation thus preventing some esophageal collapse.

Introduction

Tracheal agenesis is a rare congenital anomaly that is generally incompatible with life with a reported incidence of 0.002%. This can be associated with tracheal agenesis and management of the patient commonly accomplished via esophageal intubation with positive pressure ventilation. Overall, the prognosis is poor due to the compressibility of the esophagus as the primary airway and the other community-associated anomalies (cardiac and central nervous system malformations). Here we describe a case of tracheal agenesis, the initial management and diagnosis, and review the literature on novel approaches to treatment and tracheal reconstruction.

Case Report

A 32-week, 1700 gram male infant was born via spontaneous vaginal delivery to a 35 year old gravida 2 para 2 Korean female. The mother was noted to have a nasal deformity, minimal signs of polyhydramnios on prenatal ultrasound. Birth the patient had poor respiratory effort with cyanosis and no cry and APGAR scores of 3 and 4 at 1 and 5 minutes respectively. Also identified at birth was an imperforate anus, left polydactyly with an extra thumb, low set ears and a heart murmur. Initial attempt at bag ventilation failed and tracheal intubation was attempted. Under direct visualization the ET tube was placed between the vocal cords and then retroflexed. Nasotracheal intubation was achieved and an emergent tracheostomy was performed at the bedside. A midthoracic tracheostomy was identified and the rigid ET tube was palpable. No cartilaginous rings were identified, however the firm circular cartilage was palpated above. The presumed airway was entered and a 3.0 neonatal tracheostomy tube was placed. Immediately ventilation improved with saturations ranging from the 70-80s to the 90s, bilateral breath sounds were heard and chest rise was seen. Flexible laryngoscopy through the trachea revealed the tracheostomy tube to be lying within a soft tissue lumen with no identifiable carina or tracheal rings. Direct laryngoscopy showed hypomobility of the left true vocal fold and the flexible scope could not be passed through the subglottis which had no obvious lumen.

An OGT was easily passed into the esophagus under direct visualization, however the patient began to desaturate and the OGT was removed with subsequent recovery of his saturation. Post-operative chest x-ray revealed gastric dilation, distal esophageal dilation, and elevated lung spaces.

Discussion

At birth the patient had poor respiratory effort with cyanosis and no cry. The classical presentation of a baby, usually less than 2,500 grams, with respiratory difficulty, no audible cry and difficult intubation despite clear visualization of the vocal cords should be recognized by all the health professionals that are most likely to be present in the first hours of the baby being born i.e. anesthesiologists, obstetricians, neonatologists and otolaryngologists. If the classic presentation is recognized, an esophageal intubation should be attempted. Due to the compressibility of the esophagus, ventilation could still be a challenge. In our case, after nasal intubation, despite having some degree of ventilation (SpO2 of 70%), the saturation did not increase over this level until the placement of the tracheostomy tube. The tracheostomy tube is more rigid and allows for higher pressure ventilation thus preventing some esophageal collapse.

Generally, this has been a fatal condition, though there are several reports of surgical strategies that have led to children living in early childhood. The longest surviving infant born with tracheal agenesis was reconstructed by Soh and colleagues and lived to six years of age.

The patient had a Floyd I type tracheal agenesis. They did a double lumen tube and placed a nasopharyngeal stent and an endotracheal tube through the distal stoma. The patient did not require ventilatory support. Unfortunately, the patient had an acute respiratory distress syndrome secondary to tracheal agenesis. The case of Floyd type II tracheal agenesis was managed with a tube placement into the esophagus with later colonic graft reconstruction of the esophagus allowing the patient to breathe room air, speak with oclusion of the tube orifice and eat small amounts of formula.

These reconstruction techniques are unique, labor intensive and require multiple surgeries and prolonged hospitalization. In addition, they do not allow for normal speech or eating. New research and applications of tissue engineering may offer an alternative option for these cases in the future.

Recently a comprehensive review by Lange, et al describes the tissue engineering technology and proposed use in laryngotracheal reconstruction. The tissue can be used to replace a short segment of the trachea in cases of tracheal agenesis. The tissue can be used to replace a short segment of the trachea in cases of tracheal agenesis, thus allowing a longer length of tissue to be engineered reconstruction of the airway, including a complete circumferential adult trachea that was made in vivo and transplanted into a 10 year old boy who had congenital tracheal stenosis. The construct is made by providing a scaffold, either synthetic or decellularized allograft, on which epithelial stem cells can grow. The success to date shows great promise for future reconstruction, however, there is still a great deal of research to be done.

Conclusion

A high degree of suspicion is necessary to identify this rare congenital anomaly. The classic presentation of a neonate in respiratory distress often accompanied by multiple organ system anomalies should raise suspicion and an esophageal intubation should be attempted if direct laryngoscopy with clear visualization of the cords proves unproducible. At this time there is no satisfactory replacement for the tracheal agenesis. The success to date shows great promise for future reconstruction, however, there is still a great deal of research to be done.

References

6. Payne was the first to describe tracheal agenesis in 1900. J Pediatr. 1900;14:568.

Figure 1. Plain radiograph of patient. A) Lateral view with posteriorly displaced tracheostomy tube (yellow arrow), B) Anteroposterior view after isovue injected through tracheostoma adjacent to tracheostomy tube showing evidence of contrast in distal esophagus (green arrow) with a bronchogram (blue arrow).

Figure 2. Floyd’s Classification from Haben CM, Rappaport JM, Clarke KD. Tracheal Agenesis. J Am Coll Surg. 2002;194:217-222.