

CASE REPORT

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Early ligation of the gastroesophageal junction to manage respiratory instability in tracheal agenesis: A case report

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ABSTRACT

Introduction: Tracheal agenesis (TA) is an exceedingly rare life-threatening congenital malformation. It can present in isolation, as part of congenital syndromes, or in the setting of chromosomal abnormalities and is associated with recognized risk factors such as maternal diabetes. An associated tracheoesophageal fistula (TEF) can allow for ventilation at birth via the esophagus. However, mortality among patients with TA remains high, even with prompt surgical intervention.

Case Report: We present the case of a male infant who had delayed diagnosis of TA. The patient underwent emergent exploratory laparotomy and ligation of the gastroesophageal junction to temporize his respiratory instability but ultimately was transitioned to comfort focused care following multidisciplinary discussion with his family.

Conclusion: This case highlights the importance of early recognition of TA. Temporizing measures, including surgical ligation of the gastroesophageal junction, can allow time for more definitive treatments.

Keywords: Gastroesophageal ligation, Pediatric surgery, Tracheal agenesis

How to cite this article

Amann J, McRae JLLH, Kashtan M, Radulescu A, Mejaddam A. Early ligation of the gastroesophageal junction to manage respiratory instability in tracheal agenesis: A case report. J Case Rep Images Surg 2025;11(2):5–9.

Article ID: 100159Z12JA2025

doi: 10.5348/100159Z12JA2025CR

INTRODUCTION

Tracheal agenesis (TA) is a rare condition, presenting 1 in 50,000 live births and affects males twice as often as females [1]. It carries a grim prognosis with few children making it past the age of 4 and the majority dying within the first week of life [2]. The anatomic malformation of the trachea lies on a spectrum from partially developed to completely absent, with or without the presence of a tracheoesophageal fistula (TEF) [3]. The absence of a patent TEF is not compatible with life after parturition.

CASE REPORT

The patient was a male born at 37 weeks gestation with no prenatal care until approximately 28 weeks gestation due to history of irregular menstrual cycles. The prenatal period was notable for severe polyhydramnios and maternal diabetes discovered at 36 weeks gestation. Prenatal echocardiogram revealed patent ductus arteriosus and double outlet right ventricle (DORV). Upon delivery via scheduled cesarean section, there was no spontaneous crying and cord clamping was delayed for 1 minute per institutional protocol. He was placed on positive pressure ventilation (PPV) due to absent respiratory effort. Despite deep oral suctioning of thick secretions, there was no improvement in oxygenation,

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Received: 29 March 2025

Accepted: 28 June 2025

Published: 31 July 2025

and he had sustained bradycardia. After two failed intubation attempts with a 3.5 and 3.0 endotracheal tube (ETT), the third attempt with a 3.0 uncuffed ETT was deemed successful and confirmed with appropriate end tidal carbon dioxide (CO_2). At this time, it was not appreciated that the ETT was in the esophagus. The ETT was secured at the mouth at 12 minutes of life and fraction of inspired oxygen (FiO_2) was maintained at 75–85%. Appearance, pulse, grimace, activity, and respiration (APGAR) scores were 2, 2, and 8 at 1, 5, and 10 minutes, respectively. Initial chest X-ray (CXR) demonstrated the ETT tip overlying what was thought to be the mid thoracic trachea, with a prominent cardiac silhouette and diffuse pulmonary interstitial prominence potentially associated with retained fluid, edema or infection (Figure 1).

Physical assessment revealed macrocephaly, cyanosis, and poor tone. An umbilical artery catheter and orogastric tube (OGT) were placed. Transthoracic echocardiogram on day of life (DOL) 1 revealed Tetralogy of Fallot with pulmonary atresia, large inlet type ventricular septal defect (VSD) with overriding aorta, and moderate sized patent ductus arteriosus. Brain magnetic resonance imaging (MRI) and head ultrasound on DOL 2 demonstrated stenosis of the cerebral aqueduct with concurrent lateral and third ventricle ventriculomegaly with absence of the septum pellucidum. A large air leak was noted on DOL 2 and an attempt to upsize the ETT was attempted but was unsuccessful, with poor visualization due to many bubbly secretions. The air leak was subsequently tolerated as the patient was still ventilating and oxygenating adequately. On DOL 3 the patient was taken to the operating room for a ventriculostomy to manage the ongoing hydrocephalus.

Appreciating the air leak, the anesthesia team attempted again to replace the ETT with a cuffed tube, however this was unsuccessful, and the patient immediately began to decompensate. A laryngeal mask airway (LMA) was placed and the Otolaryngology (ENT) service consulted intraoperatively. Emergent laryngoscopy demonstrated that the trachea ended in a blind pouch shortly after the false vocal folds (Figure 2). Subsequent esophagoscopy demonstrated a TEF (Figure 3). The Pediatric Surgery service was then consulted emergently. An exploratory laparotomy was performed and the gastroesophageal junction was occluded with a Rummel tourniquet, resulting in significant improvement of oxygenation and ventilation through the esophageal ETT. Two metal clips were placed across the gastroesophageal junction and a negative pressure temporary abdominal closure dressing was placed to allow for future intervention.

Over the next several days, a multidisciplinary team met with the family to discuss treatments and potential plans of care including surgical reconstruction. Following these discussions, comfort-focused care was chosen by family and the patient expired on DOL 7.



Figure 1: Chest X-ray demonstrating initial placement of ETT on first day of life.

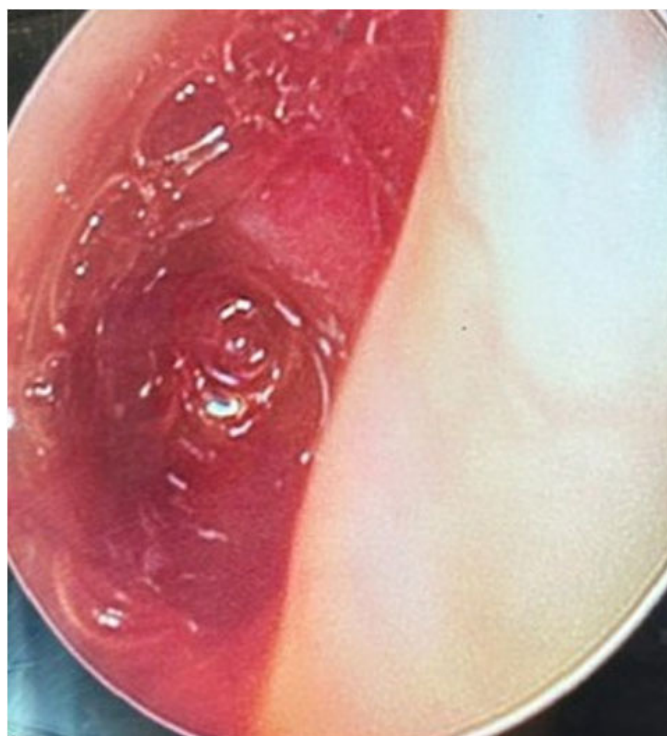


Figure 2: Endoscopic image demonstrating the proximal trachea terminating in blind pouch.

DISCUSSION

Tracheal agenesis (TA) is an exceedingly rare congenital malformation, with fewer than 200 reported cases [2]. It is difficult to diagnose prenatally, with the vast majority of cases being discovered shortly after parturition or upon autopsy. Only seven cases of TA



Figure 3: Endoscopic image demonstrating a distal esophageal fistula communicating with airway.

in the reported literature were diagnosed prenatally [4]. According to Çelik et al., the incidence of TA is approximately 1 in 50,000 newborns [1].

When identified prenatally, TA often presents similar to congenital high airway obstruction syndrome (CHAOS) with large echogenic lungs, flattened diaphragm and lack of flow in the trachea [5]. One diagnostic tool that has been promising in prenatal diagnosis of TA is amniocentesis assessing for phospholipids [6]. Phospholipids are primarily secreted into the amniotic fluid via the respiratory tract, and therefore, an absence of phospholipids upon amniocentesis can be an indicator of an underdeveloped respiratory tract, possible TA [6]. Another helpful tool in the workup of TA is prenatal ultrasonography demonstrating polyhydramnios, which may indicate obstruction of the airway or esophagus [1, 2, 4–6]. TA typically presents at birth with a lack of audible cry, respiratory distress, and multiple failed intubation attempts [1, 2, 4–8]. An APGAR score below 7 at 5 minutes is a clinical sign often seen in TA [4]. Other possible postnatal signs that can aid in the diagnosis of TA after parturition include collapsed lungs on chest X-ray, severe cyanosis, absent breath sounds on auscultation, and minimal to absent chest rise [1, 2]. Postnatal diagnosis of TA is often made difficult, especially in the context of a lack of prenatal care. Conventional chest X-rays are also non-specific and often do not provide enough contrast or differentiation to identify congenital airway defects. Bronchoscopy is the gold standard in identifying cases of TA post-partum. TA is part of multiple syndromes,

including TACRD (Tracheal agenesis, Congenital cardiac defects, Radial ray anomalies, and Duodenal atresia) and VACTERL (Vertebral defects, Anal atresia, Cardiac defects, Tracheoesophageal fistula, Esophageal atresia, Renal dysplasia, and Limb abnormalities) [1, 3, 9]. Other associated congenital anomalies include annular pancreas, umbilical cord defects, neurological conditions including ventriculomegaly and hydrocephalus, and specific cardiac defects including an atrioventricular septal defect and Tetralogy of Fallot [3].

This case demonstrated the importance of a comprehensive anomaly workup including echocardiography, renal ultrasound, and head and spine imaging. Genetic testing as well as association screening for associations like TACRD and VACTERL can clarify a patient's unique presentation [9]. Airway anomalies must be prioritized first due to their danger with misdiagnosis [1]. Incorporating a multidisciplinary team effort is essential to addressing other associated conditions and syndromes. This team-based approach helps inform the scope of anomalies, assess overall prognosis, and support shared decision-making with the family.

There are several different classification systems employed for TA, the most widely used of which was first described by Dr. John Floyd in 1962. Under Floyd's classification scheme, agenesis of the proximal trachea with a short segment of normal distal trachea, carina, and bronchi is Type I. Type II is agenesis of the entire trachea with an intact carina, and Type III is atresia of entire trachea and carina with bronchi originating individually from the esophagus [10]. In the case of our patient, Type II TA was suspected due to the trachea appearing to bifurcate at the carina immediately at its junction with the esophagus. This is the most commonly reported type [5].

No specific teratogenic medications, toxins, or vitamins have been specifically linked to the development of TA, but recent research has proposed maternal diabetes as a potential contributor to the development of airway anomalies [11]. Several different key molecular signaling events during embryogenesis have been linked to the development of TA, in which the ventral foregut fails to develop into a trachea with a separate esophagus. This absence of ventral patterning has been linked to the abnormal/absent expression of Nkx2.1 [9]. It was discovered after postmortem genetic testing that our patient has a de novo sequence variant c.424CT in the IQGAP1 gene, which has been linked to premature protein termination. Though it is thought that this variant may be pathogenic, at this time, the clinical significance of this mutation is unclear. There have been reported cases of de novo and inherited variants in the IQGAP1 gene in patients with congenital heart disease, including Tetralogy of Fallot and transposition of the great arteries [12, 13]. Our patient was negative for any microdeletion or microduplication syndromes, or autosomal and sex chromosome aneuploidy.

Due to the blind ending trachea in TA, endotracheal intubation is impossible. Bag and mask ventilation has been proven to be successful in some cases where the TEF is large enough to ventilate the lungs [14]. Esophageal intubation, when a TEF is present, has been shown to be one of the most effective methods in permitting adequate oxygenation to infants born with TA [1]. As in our case, the esophagus can be mistakenly intubated leading to the false assurance that a patent airway via the trachea has been established, which can delay or prevent the correct diagnosis of TA. Ventilating the esophagus usually causes distension of the stomach [8], potentially necessitating ligation of the gastroesophageal junction and associated gastrostomy until definitive surgical repair [5].

Once diagnosis of TA has been made with laryngoscopy, cross-sectional imaging can be utilized to better delineate the anatomic defect. Cristallo et al. utilized computed tomography (CT) to demonstrate an absence of tracheal air column [4]. Coleman et al. utilized thoracic magnetic resonance imaging (MRI) to reveal TA with a blind pouch just below the vocal cords with presence of a distal TEF [7].

The surgical reconstruction of infants born with TA is the only viable treatment modality and is highly dependent on the type and severity of agenesis [14]. This multi-faceted surgical procedure requires deliberate planning. A more recent case of Type I TA demonstrated the successful surgical reconstruction of the airway and implementation of a 3D-printed external bioresorbable splint for external esophageal airway support [15]. In the literature, there have been only 7 survivors with TA who underwent surgical reconstruction and 5 of them received some form of external splint to prevent airway collapse [15]. This demonstrates the necessity of adequate airway support when considering the surgical reconstruction of patients with TA and the extreme difficulty in managing these patients.

Tracheal reconstruction is not ideal in an emergent situation, where trying to establish and maintain a proper airway is the chief concern. As was mentioned previously, esophageal intubation is one of the most effective ways to ventilate infants with TA and patent TEF at parturition, however it comes with the associated dilation of the stomach with air [8]. In our case, an exploratory laparotomy was performed to ligate the gastroesophageal junction (GEJ), thus avoiding insufflation of the stomach and improving lung ventilation.

CONCLUSION

Tracheal agenesis is a rare congenital malformation that typically presents with absence of crying, respiratory failure, low APGAR scores, and multiple failed intubation attempts. Our patient presented with the aforementioned characteristics as well as concomitant Tetralogy of Fallot, pulmonary atresia and obstructive hydrocephalus. In emergent cases where there is loss of airway in patients with suspected TA with TEF, esophageal intubation with

subsequent ligation of the GEJ should be considered as a temporizing measure to definitive intervention.

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Author Contributions

Jacob Amann – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Joyce JLH McRae – Conception of the work, Design of the work, Analysis of data, Drafting the work, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Mark Kashtan – Acquisition of data, Analysis of data, Interpretation of data, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Andrei Radulescu – Conception of the work, Design of the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Ali Mejaddam – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation

of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Guarantor of Submission

The corresponding author is the guarantor of submission.

Source of Support

None.

Consent Statement

Written informed consent was obtained from the patient for publication of this article.

Conflict of Interest

Authors declare no conflict of interest.

Data Availability

All relevant data are within the paper and its Supporting Information files.

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