CASE REPORT

Tracheal agenesis: A rare but fatal congenital anomaly

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ABSTRACT: In this report we describe a newborn with a rare case of Type II tracheal agenesis and bronchoesophageal fistula. Polyhydramnios and suspected esophageal atresia were identified during routine pre-natal ultrasound screening. Upon delivery, rigid bronchoscopy, esophagoscopy, and intraoperative fluoroscopy were performed, where both bronchi and the carina showed unusual horizontal orientation making it difficult to identify the fistula. However, a post mortem CT confirmed the diagnosis of an isolated Type II tracheal agenesis with bronchoesophageal fistula.

INTRODUCTION

Tracheal agenesis is a rare and lethal congenital anomaly, where a complete interruption or absence of the trachea is present. Since it was initially described in 1900, few cases have been published worldwide (1). The prevalence of tracheal agenesis is less than 1:50,000 with a male to female ratio of 2:1. In general, 52% of cases are associated with premature delivery and approximately half of the cases are associated with polyhydramnios (2).

Floyd et al. classified tracheal agenesis into three widely accepted anatomical subtypes (3). Type I is characterized by agenesis of the proximal trachea and by the presence of a distal tracheoesophageal fistula, whereas type II is defined by a complete absence of the trachea and by the presence of normal bifurcating bronchi. In type III, the two main bronchi arise independently from the esophagus (3).

Isolated Type II tracheal agenesis with fistula is rare, and up to 94% of cases are associated with other congenital abnormalities. Newborns with Type II tracheal agenesis usually present with polyhydramnios, respiratory distress, aphonia, cyanosis, and an inability to undergo endotracheal intubation. Currently, type II tracheal agenesis accounts for approximately 50-60% of all cases. At present, the etiology of type II tracheal agenesis is unknown and no significant karyotype exists (4).

In general, prenatal diagnoses can only be made via ultrasonography and the outcome is poor (5,6). This case report depicts an unusual situation where Type II tracheal agenesis with esophageal fistula was diagnosed independently of any other congenital anomalies.

CASE REPORT

Our patient was born to a 31-year-old G2P1A0 mother and a non-consanguineous father. Prenatal ultrasound at 25, 29, and 32 weeks showed polyhydramnios and suspected esophageal atresia. Amniocentesis reductions were performed three times during the pregnancy to reduce the volume of amniotic fluid. Aside from these findings, the pregnancy was without other known complications. The patient was delivered at 35 1/7 weeks gestation by caesarean section owing to fetal bradycardia and the suspected diagnosis

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of esophageal atresia. The baby made no audible cry. Its birth weight was 1850gm, indicative of intrauterine growth restriction (IUGR). Respiratory distress developed immediately following birth and attempts at intubation with a 2.5 mm endo-tracheal tube (ETT) failed due to the blockage below the vocal folds.

Examination showed a hypoxic newborn with a PO_2 of 63, with a blood pH of 6.8 and a PCO_2 of 135. Mask ventilation was also difficult, but an O_2 saturation of 95% was achieved following administration of 75% FiO_2.

Once stabilized, the perinatal patient was immediately transferred to the operating room of a tertiary pediatric medical center. A second intubation using a 2.5 mm ETT failed due to subglottic obstruction. Rigid bronchoscopy was performed and revealed a blind pouch below the vocal folds (fig.1).

Tracheostomy was attempted but was unsuccessful as no cervical trachea was palpable below the cricoid cartilage. Intraoperative fluoroscopy of the trachea also confirmed the subglottic stenosis and absence of the lower cervical and thoracic trachea. The presence of a bronchoesophageal fistula was identified with both bronchi originating from a flattened carina. The bronchi and carina exhibited unusual horizontal orientation and the carina demonstrated a straight continuous roof with no tracheal remnants (fig.2).

Based on these imaging studies, isolated Type II tracheal agenesis with bronchioesophageal fistula was diagnosed. The patient died within 5 hours of birth. A post-mortem CT scan revealed a 7 mm remnant segment of cervical trachea (with no lumen) running in the craniocaudal plane and confirmed the complete absence of the lower cervical and thoracic trachea. The carina originated from the esophagus and joined to the right and left main bronchi.

DISCUSSION

Floyd et al. defined Type II tracheal agenesis as complete absence of the trachea with the presence of normal bifurcating main bronchi (3). 94% of cases are associated with other congenital defects including congenital cardiac, genitourinary, gastro-intestinal, pulmonary, CNS, and musculoskeletal anomalies (3,6,7).

While the embryological mechanisms behind Type II tracheal agenesis remain controversial, it has been suggested that tracheal agenesis is a result of the abortion of the lung bud outgrowth and delayed formation of the bronchi and lungs via remnant primordial mesenchyme which often attaches to the esophagus (7). Also, while no significant genetic karyotype has been found to correlate with tracheal agenesis, homozygous Shh-null mutant mice show foregut defects similar to those seen in tracheal agenesis (8).

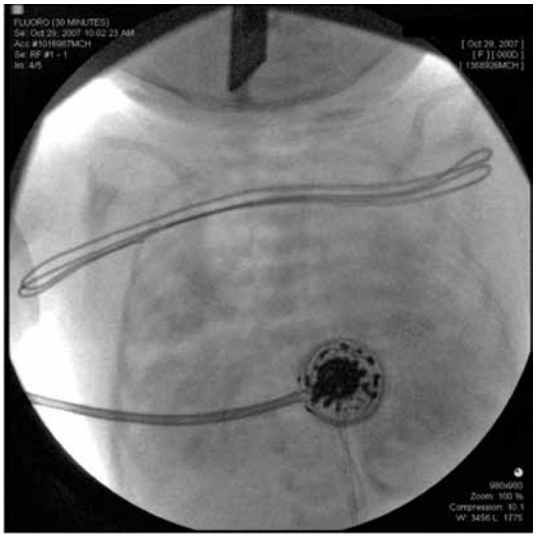

Figure 1: Notice the scope in the subglottic segment demonstrating absent cervical and thoracic trachea.

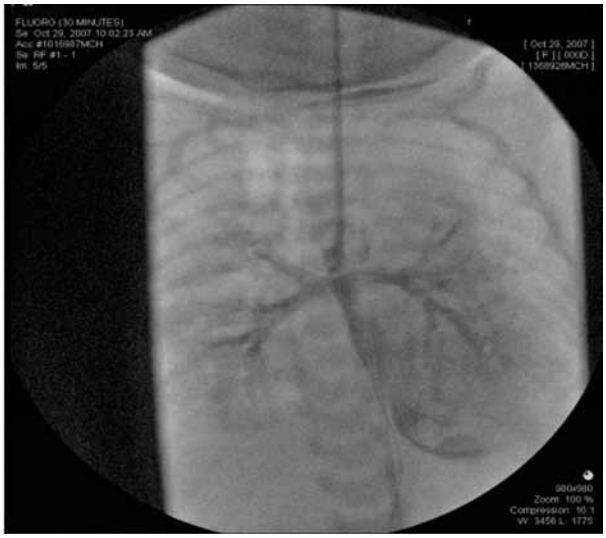

Figure 2: Note the unusual horizontal orientation of R/L main bronchi and carina (with contrast in the esophagus and bronchial tree) visualized using fluoroscopy.

Tracheal agenesis should be included in the differential diagnosis when the following clinical signs are manifested: neonate with a history of polyhydramnios, absence of an audible cry at birth, failure to intubate beyond the vocal folds, and respiratory distress (9).

In patients with tracheal agenesis, surgical correction has been proposed as a corrective measure (6). To date, only one patient with Type II tracheal agenesis has survived beyond the neonatal period. However, that surviving patient had a proximal tracheoesophageal fistula in addition to a bronchoesophageal fistula, which allowed the successful establishment of a permanent airway by performing a tracheotomy and inserting a long T tube to create a patent airway (10). While temporary management can include the insertion of an esophageal tube, depending on the extent of the fistula, infants with a complete absence of the trachea tend to die within hours of birth, as a permanent airway cannot be created. Mask ventilation can also be used to assist in temporary prolongation of life. However, there is no established medical protocol for life conservation in isolated Type II complete tracheal agenesis. Currently, this anomaly is incompatible with life and future hopes for survival will depend on surgical developments.

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