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Tracheal agenesis and esophageal atresia with proximal and distal bronchoesophageal fistulas

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Abstract Tracheal agenesis (TA) is an extremely rare, typically fatal congenital tracheal malformation. Lack of prenatal symptoms and emergent presentation usually lead to a failure to arrive at the correct diagnosis and manage the airway properly before the onset of irreversible cerebral anoxia. Esophageal atresia (EA) encompasses a group of congenital anomalies comprising an interruption of the continuity of the esophagus with or without a persistent communication with the trachea. In 86% of cases, there is a distal tracheoesophageal fistula (TEF); in 7%, there is no fistulous connection, whereas in 4%, there is a TEF without atresia. We report the case of an infant born with TA and EA with proximal and distal bronchoesophageal fistulas. During 3 consecutive antenatal ultrasound examinations, there had been polyhydramniosis, difficulty visualizing the stomach, and dilatation of proximal esophagus, leading to a presumptive diagnosis of EA. The clinical presentation, embryology, classification, and surgical management are discussed.

Tracheal agenesis (TA) is an uncommon congenital malformation with an extremely high mortality rate. This malformation manifests after delivery as severe respiratory distress, cyanosis and lack of crying, and impossibility of endotracheal respiratory support. This anomaly is usually associated with cardiovascular, respiratory, and gastrointestinal malformations. Only a high index of suspicion and early surgical management will prevent death. An early diagnosis may also help the parents and the medical team to take appropriate decisions. Currently, there is no effective therapy that guarantees long-term survival [1].

Esophageal atresia (EA) occurs in 1 in 2500 live births. Infants with EA are unable to swallow saliva and are noted to have excessive salivation requiring repeated suctioning. Associated anomalies occur in 50% of cases, most involving one or more of the VACTERL (vertebral, anorectal, cardiac, tracheoesophageal, renal, and limb defects) association. The etiology is largely unknown and is likely to be multifactorial; however, various clues have been uncovered in animal experiments particularly defects in the expression of the gene Sonic hedgehog (Shh). Most cases are sporadic, and the recurrence risk for siblings is 1%. The diagnosis may be suspected prenatally by a small or absent stomach bubble on antenatal ultrasound scan at around 18 weeks of gestation. The likelihood of an atresia is increased by the presence of polyhydramnios. Survival is directly related to birth weight and to the presence of a major cardiac defect. Infants weighing more than 1500 g and having no major cardiac problem should have a near 100% survival, whereas the presence of one of the risk factors reduces survival to 80% and further to 30% to 50% in the presence of both risk factors [2].

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1. Case report

A male infant was born by lower-segment cesarean delivery at 35 weeks of gestation to a 27-year-old mother. Polyhydramniosis, a dilated upper esophageal pouch and no visualization of the stomach, was found during 3 consecutive prenatal examinations by ultrasound. These findings were leading to a presumptive diagnosis of esophageal atresia were found. At delivery, the infant had respiratory distress and cyanosis that did not improve after stimulation and ventilation via mask. Tracheal intubation was impossible because of a complete obstruction at the level of vocal cords. The nasogastric catheter was pushed into the esophagus, but it did not pass through the stomach. On the other hand, this esophageal catheter could oxygenate the patient as well as having gastric distension. Patient immediately delivered premature intensive care unit. A chest radiograph confirmed position of the nasobronchial catheter and esophageal atresia.

Esophageal atresia and esophagobronchial fistula were confirmed by esophagobronchoscopy. Firstly, cervical exploration showed a TA. Then we performed a gastrostomy procedure by midline vertical abdominal incision. Because airflow from distal esophagus to stomach was observed during the operation, a high esophageal ligation procedure was performed to block airflow into the stomach and to prevent reflux of gastric content. After that, the ventilation of baby became much easier. Although cardiac arrest developed during operation, patient revived after resuscitation. In the postoperative fourth hour, he developed cardiac arrest while still on mechanical ventilation via esophageal intubation and medical cardiac support treatment. Despite all resuscitation attempts, patient could not be revived (Fig. 1). The family did not accept the autopsy.

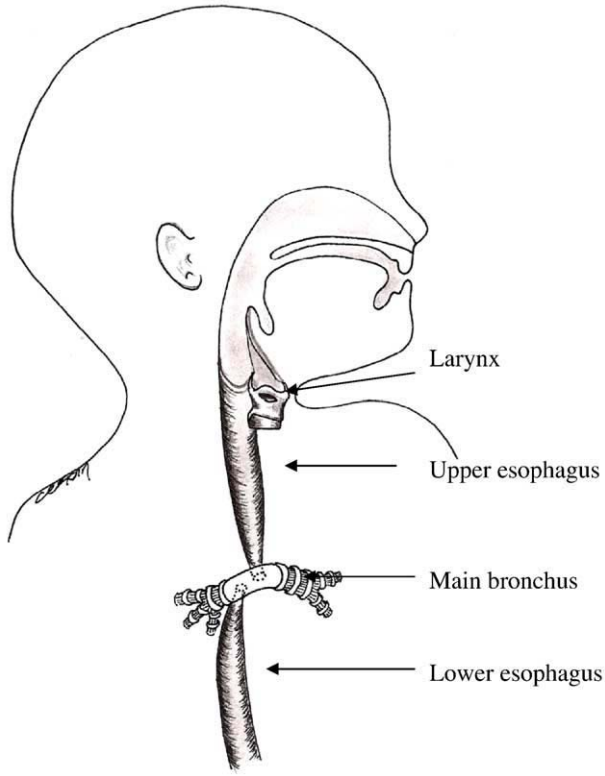

Fig. 1 Schematic appearance of tracheal anomaly in patient.

2. Discussion

Tracheal agenesis is one of the rarest congenital anomalies of the airway. It reported firstly in 1900 by Payne [3]. Fewer than 100 cases have been reported in the literature until today. The incidence of it was reported to be less than 1:50,000 with a male predominance [1,4]. It presents immediately at birth with cyanosis and respiratory distress. The infant makes no audible sounds. Numerous attempts at neonatal resuscitation with endotracheal intubation meet with failure, although the larynx and vocal cords are well visualized. The classification system of Faro [1,5] seems to have the most practical application from a surgical standpoint. Faro et al [1,5] divided tracheal atresia into categories A to G, representing various types of airway anomalies decreasing in severity, with category A representing total pulmonary atresia and category G representing tracheal stenosis. Our case fitted type B of classification of Faro.

Embryologically, TA results because of altered formation of the tracheoesophageal septum. According to Kluth et al [6], TA with fistula may result from a ventral deformation of the foregut and a concomitant dorsal dislocation of the tracheoesophageal space. In another theory, TA results when normal tracheal elongation fails to take place. The degree to which the trachea develops determines the type of TA that results. Failure of respiratory diverticula's formation leads to Faro "type A" anomaly representing total pulmonary agenesis. If the respiratory diverticulum fails to fuse, separate bronchoesophageal fistulae persist, and TA type B results. Tracheal agenesis types C and D occur if the respiratory diverticulum fuse to form the carina, but tracheal elongation does not occur. Arrested elongation of the trachea with resultant failure to fuse with the developing larynx results in TA Faro type E, F, or G. Various other abnormalities have been described in association with TA. Evans et al [7] described a pattern of malformation that includes TA, complex congenital heart anomalies, radial defects, and duodenal atresia. The underlying mechanism responsible for these heterogeneous groups of malformation remains obscure but may be related to abnormal epithelial mesenchymal interactions or a disruption of early blastogenesis. Typically, TA is fatal in the neonatal period, and the diagnosis is not made until a postmortem examination. Very few reports have demonstrated long-term survival with surgically reconstructed TA [1,8]. If surgical treatment is planned, then initial stabilization of these infants requires esophageal banding to prevent regurgitation of gastric fluid and abdominal distension owing to positive pressure mechanical ventilation [9]. Cervical esophagostomy

is necessary for salivary drainage to prevent aspiration pneumonia. Tracheostomies are generally unsuccessful in these babies [10], and insertion of a tracheostomy tube through a proximal esophagostomy into the esophagobronchial communication has been described [11]. We have chosen proximal esophageal tube insertion for ventilation, gastrostomy, and ligation of distal esophagus as an initial procedure. Furthermore, once it was discovered that the infant had TA, extracorporeal life support (ELS) or bypass could have been considered to stabilize the patient. But the baby lived only 4 hours. Our anticipation for diagnosis is EA. If TA might be suspected prenatally, we could have been ready to use the ELS or bypass.

Definitive surgical reconstruction for patients with a distal tracheal remnant continues to be challenging. Esophageal diversion followed by reconstruction with colonic interposition or gastric pull-up has already been demonstrated to be feasible [1,12]. However, at present, there is no suitable autologous tissue to assume the function of the normal trachea with its rigid cartilaginous framework. Although various types of homologous grafts such as trachea, esophagus, pericardium, and bladder as well as synthetic ones (silicone, elastane, and Dacron) have been used to reconstruct the trachea, results has been not good [1,13,14]. Cartilaginous tissue engineering may offer a solution in the future.

It is important to remember TA can be falsely diagnosed by antenatal ultrasonogram as an EA. At postnatal period, this kind of case can be mixed with laryngotracheal cleft as well. It is helpful to determine the pathologic condition by endoscopy and detecting the cartilage tissue in the biopsy specimens.

In conclusion, in early recognition and proper airway stabilization by esophageal intubation, ELS or bypass technique may allow short-term respiratory support and the prevention of acute anoxia or hypoxia in TA. However, despite early diagnosis of this anomaly, the question has yet remained whether any of the reconstructive surgical options are justified or worthwhile. More knowledge about the embryological formation of the trachea and advances in surgical reconstruction techniques are required to survive these babies in the future.

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