

Tracheal atresia with a cephalically developed lung bud

A case report

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Abstract

Rationale: Tracheal atresia (TA) involves complete or partial tracheal absence below the larynx. It involves failure of complete separation of the laryngotracheal diverticulum from the foregut at the 4th gestational week. In TA, the trachea and main bronchi generally proceed in the normal caudal directions.

Patient concerns: At the gestational age of 34 weeks and 6 days, a male baby weighing 2290 g was born via cesarean section. A brisk bag was used, and mask ventilation was performed, but was not effective. Intubation was attempted; however, the endotracheal tube did not advance below the vocal cord.

Diagnosis: Tracheal atresia.

Interventions: An emergent open neck exploration could not reveal the trachea. On computed tomography, the tracheoesophageal fistula (TEF) started from the lower esophagus just above the gastroesophageal junction. The trachea arose upward to form the main bronchus.

Outcomes: His parents signed the “Do not resuscitate” form due to poor outcome. On the 23rd day of birth, the baby expired.

Lessons: Our case involved TA with tracheoesophageal fistula forming between the trachea and main bronchi in a cephalic direction as detected on computed tomography. Further, the trachea arose from the gastroesophageal junction; thus, it does not belong to any Floyd classification. Herein, we report a TA case with a cephalically developed lung bud.

Abbreviations: AVSD = atrioventricular septal defect, DORV = double outlet right ventricle, PAPVR = partial anomalous pulmonary venous return, TA = tracheal atresia, TEF = tracheoesophageal fistula.

Keywords: cephalad, esophagus, lung bud, tracheal agenesis, tracheal atresia

1. Introduction

Tracheal atresia (TA) is a disease involving the complete or partial absence of the trachea below the larynx.^[1] Payne reported the first case of TA in 1900; since then, only approximately 150 cases were reported worldwide. The incidence was <1 in every

50,000 infants in a male-to-female ratio of 2:1.^[2] Respiratory distress at birth and failure of intubation usually occur in TA. Affected patients could breathe via a tracheoesophageal fistula (TEF) if it is present.^[1] TA is divided into 3 groups according to the part of the lower respiratory tract that initiates from the esophagus.^[3] However, the direction of the lower respiratory tract in TA is not different from that in other normal infants, that is, caudal direction. This is the first case of TA in which the lower respiratory tract arose in the cephalic position. This study was reviewed and approved by the Institutional Review Board (IRB) of Chung-Ang University. Patient has provided informed consent for publication of the case. Informed consent was confirmed by the IRB (IRB No. 1707-008-16081).

2. Case report

A 30-year-old primigravida had heart abnormalities observed on antenatal ultrasonography at the gestational age of 22 weeks in local obstetrics center and was transferred for further evaluation. She had no other history of medications or illnesses. On antenatal ultrasonography, a single umbilical artery with the following cardiac and gastrointestinal anomalies was also suspected: atrioventricular septal defect (AVSD) with partial anomalous pulmonary venous return (PAPVR) or double outlet right ventricle (DORV) and small bowel atresia. The karyotype was normal on the amniotic fluid chromosomal study. The patient was admitted twice for amnioreductions.

Editor: N/A.

Research involving Human Participants: This study was reviewed and approved by the institutional review board of Chung-Ang University (IRB No. 1707-008-16081).

The authors declare that they have no financial relationships relevant to this article to disclose and they have no competing interests.

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Medicine (2019) 98:18(e15397)

Received: 15 November 2018 / Received in final form: 25 March 2019 /

Accepted: 2 April 2019

<http://dx.doi.org/10.1097/MD.00000000000015397>

Preterm premature rupture of membranes occurred 15 hours before birth. At the gestational age of 34 weeks and 6 days, a male baby weighing 2290 g was born via cesarean section. The Apgar score was 3 at 1 minute and 5 at 5 minutes. At birth, no audible cry was heard. The chest wall was severely retracted, and the entire body was cyanotic and hypotonic. The heart rate was below 100 bpm. A brisk bag was used, and mask ventilation was performed, but was not effective. Intubation was attempted; however, the endotracheal tube did not advance below the vocal cord. A gastric tube was inserted through the vocal cords again,

but failed. An emergent open neck exploration could not reveal the trachea.

With the endotracheal tube inserted through the esophagus, the baby's spO_2 level was sustained. To avoid regurgitation of stomach juice into the airway, a gastric tube was inserted. In the neonatal intensive care unit, a mechanical ventilator was attached. The breathing sound was coarse and decreased; the heart beat was regular with systolic murmurs on the left sternal border. The abdomen was distended with normoactive bowel sounds. Single umbilical artery and preaxial polydactyly were found. His neonatal reflexes, including the moro, grasp, and sucking reflexes, were intact.

On x-ray, diffuse ground glass opacities on the entire lungs were observed. The stomach and first portion of the duodenum were distended with gas, showing a double bubble sign, which suggested a duodenal atresia. The echocardiography showed AVSD with PAPVR and DORV. On computed tomography, the TEF started from the lower esophagus just above the gastroesophageal junction. The trachea arose upward to form the main bronchus (Fig. 1A). In the transverse view, a thin membrane dissecting the trachea from the esophagus was observed in the TEF (Fig. 1B). Horseshoe kidneys were also found. An imaginary figure of TA with TEF in this case is shown in Fig. 2.

After full explanations of possible operable options but with a poor prognosis, his parents signed the "Do not resuscitate" form. All medications, except mechanical ventilator and fluid maintenance, were stopped. On the 9th day of birth, apnea was

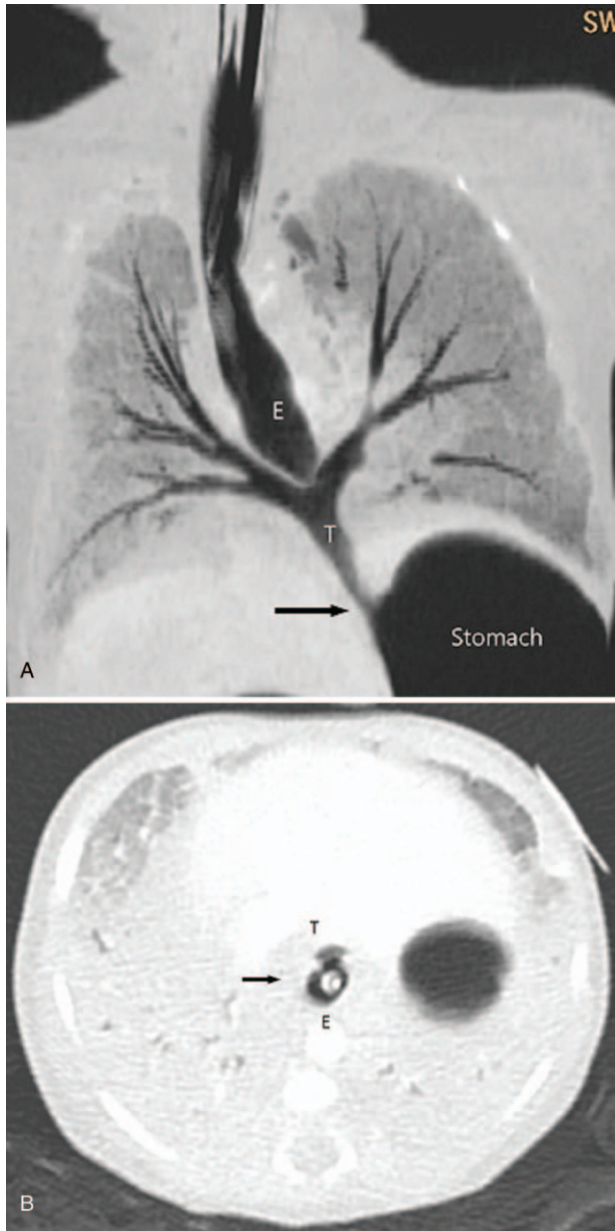


Figure 1. Chest computed tomography image of tracheal atresia; (A) (coronal view) the TEF arose from the lower esophagus just above the gastroesophageal junction. The trachea and main bronchi developed in the cephalic direction. Arrow = TEF, "E" = esophagus, "T" = trachea. (B) (Transverse view) The membranous septum dissecting the trachea and esophagus is in the TEF. The gastric tube in the esophagus is shown as a white circle. Arrow = membranous septum, "E" = esophagus, "T" = trachea, TEF = tracheoesophageal fistula.

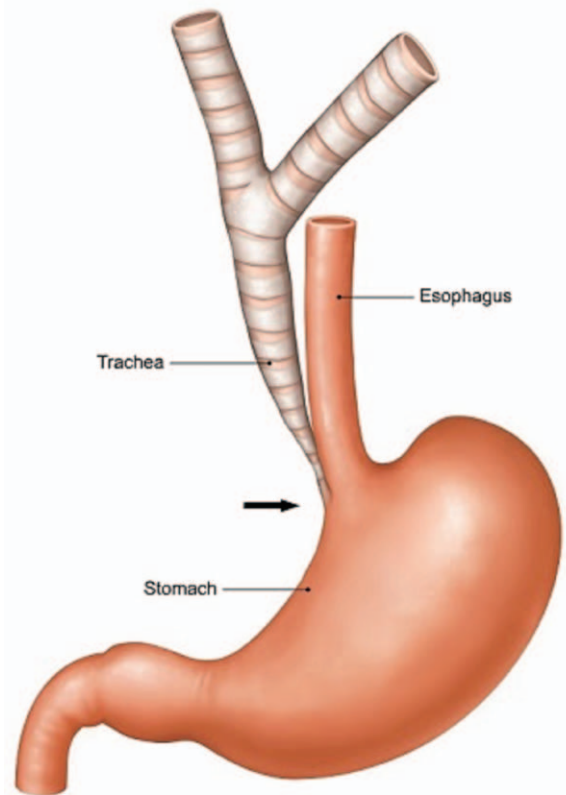


Figure 2. Imaginary figure of tracheal atresia; the trachea arises from the lower esophagus just above the gastroesophageal junction (Floyd type 1). A thin membrane is in the tracheoesophageal fistula.

observed. On the 15th day of birth, the oxygen need increased. On the 23rd day of birth, the baby expired.

3. Discussion

At the 4th gestational week, the laryngotracheal diverticulum arises from the fourth pharyngeal pouches in the caudal direction. The cranial part becomes the laryngeal epithelium, and the caudal part becomes the lower respiratory tract. The tracheoesophageal septum deepens to separate the laryngeal tube from the esophagus. The lung bud from the distal end of the laryngotracheal diverticulum becomes the bronchi and lung.^[4,5] The etiology of TA is still unclear; however, the abnormal cellular interaction between the epithelium and mesenchyme may be involved, since the respiratory tract develops from the ventral wall of the foregut in the third embryonic week.^[6] TA develops when the first outgrowth of the lung bud is aborted early during development, and the larynx or the very proximal part of the trachea stays atretic. Hedgehog signaling has been shown to be involved in this process, and exogenous factors, such as adriamycin administration, may cause tracheoesophageal malformations by interfering with this signaling pathway.^[7] Sonic hedgehog may play an important role in tracheoesophageal separation. However, the most important problem in our case was the faulty location of the lung bud. Thus, we assumed that our case was not correlated with sonic hedgehog.

We assumed that the laryngotracheal diverticulum in our case developed below the fourth pharyngeal pouches. Furthermore, the respiratory tract from the foregut formed in the cephalic direction, not in the caudal direction as usual. The lower respiratory tract from the trachea to the bronchi and lung arose upward, assuming a tree-like appearance, which is an odd shape; this is the first case of tracheal agenesis in a neonate.

Floyd et al^[3] classified TA into 3 types, depending on the part of the respiratory tract that initiates from the esophagus. In type 1, the trachea arises from the esophagus, passes to the carina, and divides into 2 main bronchi bilaterally. However, the trachea in our case arose from the gastroesophageal junction; thus, it does not belong to any Floyd classification.

We diagnosed tracheal agenesis mixed with TA. However, agenesis means congenital absence of an organ, and atresia means closure of a normal body opening or tubular structure. Tracheal agenesis develops when the first outgrowth of the lung bud is aborted early during development, and the larynx or the very proximal part of the trachea stays atretic. The paired lung primordium grows into the ventral mesenchyme, forming a bronchial tree without being connected to the larynx. Tracheal agenesis can be embryologically interpreted as a result of a defective foregut differentiation, whereas TA represents faulty development of an already differentiated anatomical struc-

ture.^[6,8] In this perspective, our case is close to TA because the trachea had a normal shape but developed upside down.

No genetic pattern has been implicated in any of the reported cases. Our case involves a problem of a directional nature. We assumed that any gene responsible for the decision regarding the direction (cephalic or caudal) may encounter problems.

Acknowledgment

The authors are grateful to Medpicture (biomedical illustration & design company) for producing the figure.

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