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Case Report

Recovery of cyanosis after esophageal intubation in a neonate with tracheal agenesis: a case report

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ABSTRACT

Background. Tracheal agenesis (TA) is a rare congenital defect that consists of a complete or partial absence of the trachea below the larynx, with or without tracheoesophageal fistula (TEF). It is a severe congenital defect with a very high mortality rate. The recommended surgical approach is esophageal ligation and gastrostomy. Despite the progress in reconstructive surgical techniques, the outcome of the anomaly is still very poor. We described a case of TA with a TEF in a female newborn with a hemivertebra, single ventricle, single atrioventricular valve, single atrium, and cardiac left isomerization.

Case. The patient, who was born at 37 weeks of age, was diagnosed with imaging methods, as the cyanosis did not improve despite being intubated many times in the delivery room; the cyanosis improved after esophageal intubation. Despite all life support treatment, the patient died on the fourth day of life. At autopsy, tracheal agenesis was diagnosed.

Conclusions. In newborns who cannot be intubated in the delivery room or whose lungs cannot be ventilated despite being intubated and whose cyanosis cannot be corrected, tracheal agenesis should be considered and ventilation with esophageal intubation should also be tried.

Key words: tracheal agenesis, newborn, heterotaxy.

Tracheal agenesis (TA) is a rare congenital defect that consists of complete or partial absence of the trachea below the larynx, with or without a concomitant tracheoesophageal fistula (TEF). This usually lethal defect is also the rarest anomaly among tracheal anomalies with an incidence of 1 per 50,000 newborns. Male patients are more likely as twice of female patients to be effected.[1]

The recommended surgical approach is esophageal ligation and gastrostomy. Despite the progress in reconstructive surgical techniques, the outcome of the anomaly is still very poor. In neonates with tracheal agenesis, severe respiratory distress and cyanosis occur just after birth and no audible cry, typically cyanosis decreases with bag-mask ventilation, but not improves or worsens by intubation. We described a case of TA with a TEF in a female newborn with vertebral anomalies (hemivertebra), congenital heart anomalies (single ventricle, single atrioventricular valve, single atrium), and cardiac left isomerization (radial ray defects).

Case Report

A female newborn was delivered from 32-year-old healthy woman by caesarean section because of fetal distress at 37 weeks and 5 days of gestation at a local hospital. The baby was the third child of healthy nonconsanguineous parents. Her brother and sister were healthy.

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At the delivery room, although spontaneous breathing movements were present, crying sounds were not heard. The newborns weight was 2400 g, her head circumference was 33 cm and Apgar scores at 1, 5, and 10 min. were 4, 5, and 5, respectively. Endotracheal intubation was performed because of severe cyanosis and bradycardia, however her oxygen saturation decreased after intubation. Therefore, bag and mask ventilation was started and her oxygen saturation increased. Tracheal agenesis and TEF were suspected and esophageal intubation was performed, oxygen saturation improved to 80% and her lungs were ventilated bilaterally. On thorax computerized tomography, a short air column at the upper part of distal trachea was visualized and disappeared at the lower part of distal trachea. As the air column was re-visualized just before the carina and main bronchia, presence of the connection between carina, main bronchi and distally esophagus were revealed.

The patient was referred to our hospital on the third day of life. Chest X-ray showed the air-filled esophagus, hemivertebra of the twelfth thoracic vertebra and bilaterally normal ventilated lungs (Fig. 1) Although the mechanical ventilation support was gradually increased, respiratory acidosis and partial carbondioxide pressure increased persistently. Echocardiography showed a functional single ventricle (single atrium, single atrioventricular valve), atrioventricular septal defect, left atrial isomerism, and patent ductus arteriosus.

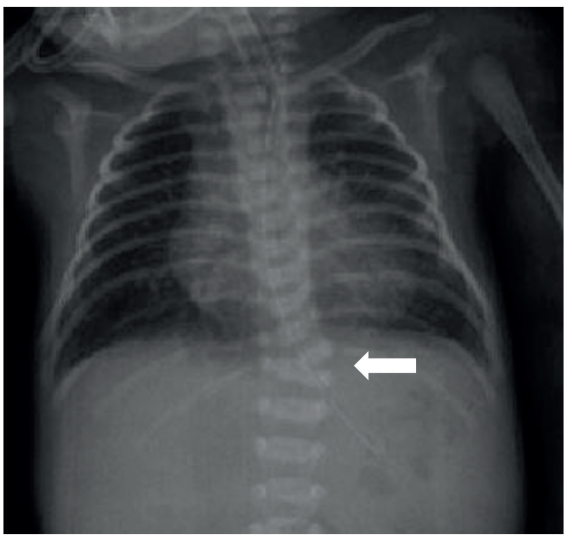

Fig. 1. Chest X-ray showed hemivertebra (white arrow) of the twelfth thoracic vertebra and bilaterally normal ventilated lungs.

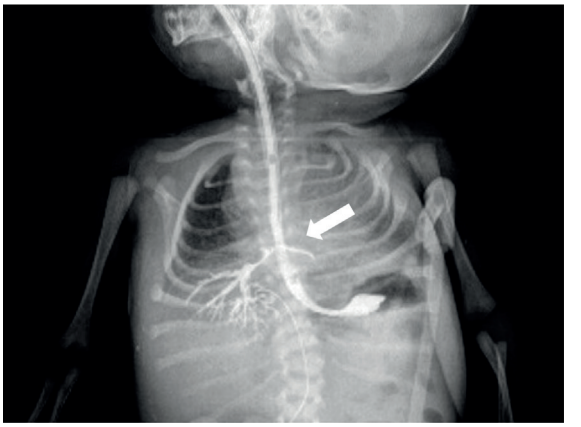

Fig. 2. The trachea was blind-ended and both main bronchi (white arrow) originated from esophagus.

Flexible laryngoscopy examination showed that the trachea was blind-ended beyond the vocal cords. Expansion of the distal esophagus was observed in esophagoscopy. In order to reduce the amount of air entering the stomach and to provide better ventilation, a Foley balloon catheter with guide wire was inserted through the esophagus into the stomach. After the balloon was inflated and pulled upward, oxygen saturation decreased more, so it was deflated and removed. It was concluded that it compressed the junction of the esophagus and main bronchia and therefore decreased the air flow to the main bronchi.

The initial surgical plan was to perform a distal esophageal ligation and gastrostomy after hemodynamic stabilization. However, despite all supportive treatment, the patient died on the fourth day of life. After the written autopsy permission was taken from the patient's family, barium sulfate was given, the trachea was blind-ended and both main bronchi originated from esophagus were demonstrated (Fig. 2).

At autopsy, TA was diagnosed. The larynx was normal but blind-ended; the pharynx continued with the esophagus which showed a connection through a TEF with two main bronchia at the bifurcation level originated from esophagus (Fig. 3). TA type was found to be type 2 according to Floyd classification and type D according to Faro classification.

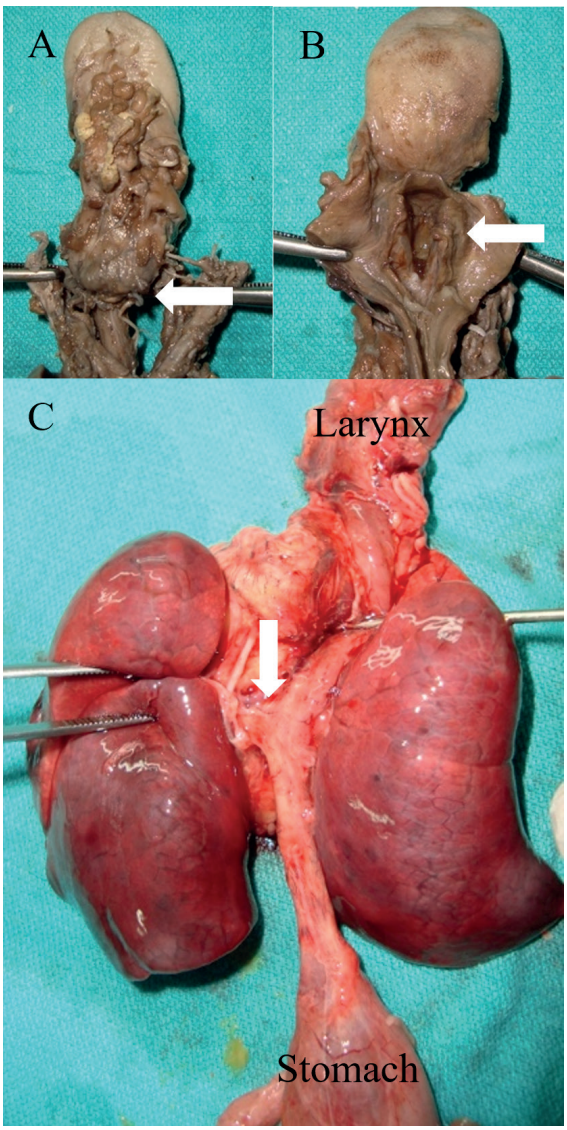

Fig. 3. A, B: At autopsy, the larynx was normal but blind-ended (white arrow). C: The pharynx continued with esophagus which showed a connection with two main bronchi at the bifurcation level through a tracheoesophageal fistula (white arrow).

The heart had single ventricle with one atrium and one atrioventricular valve. In addition, an accessory spleen and slightly dilated ureters were seen while gross examination of other organs were normal. Light microscopy revealed intra-alveolar hemorrhage, atelectasis and emphysema in lungs, and severe congestion in other organs because of hypoxia. Karyotype analysis from fibroblast cultures was 46, XX. With these results, left heterotaxy syndrome was considered in our patient.

A written informed consent was obtained from the parents for publication.

Discussion

De Groot-van der Mooren et al. reviewed in 2012, forty-nine cases of TA who have been reported since 1900.[1] Among these cases, diagnosis of TA was presumed prenatally only in five cases based on congenital high airway obstruction syndrome (CHAOS). Prenatal diagnosis in these five cases of TA was confirmed by fetal magnetic resonance imaging or detailed ultrasound examination.[1] Prenatal diagnosis is only possible in the absence of TEF with the findings of CHAOS. In other cases without prenatal diagnosis, ultrasonography showed altered amniotic fluid status, mainly polyhydramnios.[1] Our case did not have abnormal ultrasonography findings, therefore the patient could not be diagnosed prenatally.

Common clinical symptoms are respiratory distress with breathing movements without an audible cry. An interesting finding in these cases is the decreasing oxygen saturation level despite endotracheal intubation, whereas temporarily increasing oxygen saturation level when bag and mask ventilation is started. After esophageal intubation, marked abdominal distension develops, and air passes through the fistula into the lungs.[1]

Two different classifications of TA cases were made by Floyd and Faro.[2,3] While there are

types 1, 2, 3 in the Floyd classification, there are 7 types in the Faro classification as A, B, C, D, E, F, G. Floyd type 2 and Faro type D are seen most frequently. In our case, Floyd type 2 was classified as Faro type D.

Associated malformations were reported in 94% of cases with TA. Most accompanying anomalies are related to cardiovascular (64%), distal respiratory system (45-64%), gastrointestinal tract (47-50%), genitourinary tract (35-49%), musculoskeletal (19-38%), and nervous system (7%).[1,4,5] Associated malformations can be a part of the VACTERL association (Vertebral defects, Anal atresia, Cardiovascular defects, Tracheoesophageal fistula and/or Esophageal atresia, Renal defects, and Limb defects) or TARCD association (Tracheal Agenesis/Atresia, Radial ray defects, Complex congenital cardiac abnormalities, and Duodenal atresia).[4-6] In a case series including 6 patients with TA, one patient had gastrointestinal, genitourinary, cardiac anomalies and hemivertebra.[7] In our case, hemivertebra and congenital heart anomaly were accompanied by TA.

It was reported that chromosomal analysis was done in 18 cases of TA.[1] Abnormal karyotype was detected in only two cases; mos, 47, XY+mar(43,3)/46XY(56,5) and 5q11.2 deletion.[8,9] In our case, there were no apparent dysmorphic features and karyotype analysis from fibroblast cultures was 46,XX.

Radiographic findings may show posterior location of the endoctracheal tube and absence of the tracheal shadow.[1] Computerized tomography is the preferred imaging method due to giving the best and the fastest results for delineating the anatomy.[10] Radiologic imaging studies with contrast media may demonstrate a blind laryngeal/tracheal end and a TEF.[1]

The primary management includes esophageal ventilation and hemodynamic stabilization, definition of the TA with radiographic and endoscopic evaluation, followed by the initial surgical approach of esophageal ligation at the site distal to the TEF and gastrostomy. Esophagus could be used as a pseudo-trachea through this surgical procedure. Double barrel esophagostomy is to be recommended as the distal end for intubation and the proximal end for salivary drainage. An external esophageal stent can be used to avoid the esophageal collapse. Gastrostomy should be performed for feeding as a next therapeutic step. Reconstruction with small intestine, colon interposition, or gastric pull-up has been demonstrated to be feasible and compatible with survival.[11-13] Because infants with TA died shortly after birth due to severe asphyxia, surgery could not be performed in these cases. The largest review in the literature, comprising 49 cases of TA, reported that tracheostomy had been attempted in 38% of the cases to explore the options of surgical reconstruction.[1]

Unfortunately, TA has a very high mortality rate. In the largest case series of TA, mortality was high, as 34 of 40 (85%) children died within 2 days.[13,14] Despite improvements in surgical management, it was reported that only two children were still alive at 10 months and 4 years, respectively.[13,14] Soh et al.[15] reported in 1999 that among the reported cases the longest living case of TA lived up to the age of 6 years and 10 months through tracheal reconstructive surgery.[15] In 1994, a case reaching the age of 4 years with TA, proximal TEF and bronchioesophageal fistula was reported.[16] After tracheostomy was performed and long T-tube was placed, the patient was discharged from the hospital.[16] But, most of the cases of TA do not have a favorable anatomy for tracheostomy, therefore tracheostomy and T-tube do not work in these patients.

If TA is prenatally suspected, ex-utero intra-partum therapy (EXIT) should be planned.[17] EXIT procedure means providing and assuring the continuity of upper airway by tracheostomy just before clamping the umbilical cord in the delivery room.[17]

For effective tracheal repairment, special materials compatible with children's growth which are homolog tissues such as pericardium, esophagus, bladder or synthetic materials such as silicon, dacron, elastane need to be produced.[18,19] Unfortunately, biomedical technology has not yet been developed sufficiently all over the world today. Despite the progress in reconstructive surgical techniques, outcome of the TA is still very poor.

In conclusion, in newborns who have respiratory distress with breathing movements without appropriate air entry, no audible cry, and failed endotracheal intubation or whose cyanosis cannot be corrected in the delivery room, tracheal agenesis should be considered; and ventilation with esophageal intubation should also be tried.

Ethical approval

A written informed consent was obtained from the parents for the publication.

Author contribution

The authors confirm contribution to the paper as follows: case report conception and design: HTÇ, MŞA, DB, MY; data collection: HTÇ, MŞA, DB; analysis and interpretation of results: HTÇ, EŞY, MH, AES, KŞG, MY; draft manuscript preparation: HTÇ, MŞA, EŞY, MY. All authors reviewed the results and approved the final version of the manuscript.

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Conflict of interest

The authors declare that there is no conflict of interest.

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