Tracheal Agenesis : A Case Report

PAKAPHAN KIATCHOOSAKUN, MD*,
SUKANYA TAKSAPHAN, MD*,
BENJAPORN NITINAVAKARN, MD**,
JUNYA JIRAPRADITTHA, MD*,
JAMAREE TEERATAKULPISARN, MD*,
CHERDCHAI TANTISIRIN, MD***

Abstract

Tracheal agenesis is a rare congenital anomaly and typically has fatal consequences. Associated congenital malformations are present in 90 per cent of cases, most frequently affecting the cardiovascular or gastrointestinal systems and the genitourinary tract. Affected infants lack prenatal symptoms and usually present with severe respiratory distress, absence of audible crying and difficult or impossible endotracheal intubation, leading to failed airway management and irreversible cerebral hypoxia. The authors report an infant with tracheal agenesis who presented with respiratory failure after birth. The clinical features, embryology and classification schemes are presented in the hope of increasing awareness, thus making earlier diagnosis possible and thereby improving survival.

Key word : Tracheal Agenesis, Congenital Malformation

KIATCHOOSAKUN P, JIRAPRADITTHA J, TAKSAPHAN S, TEERATAKULPISARN J, NITINAVAKARN B, TANTISIRIN C
J Med Assoc Thai 2003; 86: 1086-1090

Tracheal agenesis is a rare, usually fatal, malformation often associated with other congenital anomalies. Various etiologies have been proposed but not convincingly demonstrated. Tracheal agenesis should be suspected in newborns with severe respiratory distress, absence of audible crying and difficult endotracheal intubation. Esophageal intubation can improve the respiratory condition if a tracheo-

* Department of Pediatrics,
** Department of Radiology,
*** Department of Surgery, Faculty of Medicine, Khon Kaen University, Khon Kaen 40002, Thailand.

esophageal fistula is present. Endoscopic laryngoscopy, esophagoscopy, contrast study *via* esophageal tube and CT scan can provide information on tracheal status. However, delayed or failed diagnosis of tracheal agenesis will result in cerebral anoxic damage since an adequate airway has not been established. Notwithstanding early recognition and temporary ventilatory support, most surgical reconstruction attempts fail. The authors describe a case of tracheal agenesis with a review of associated literature.

CASE REPORT

A male infant was born spontaneously to a healthy, 25-year-old mother after an uncomplicated 41-week pregnancy. The Apgar scores were 5 and 7 at 1 and 5 minutes, respectively. The child's birth weight was 2700 g. The infant was cyanotic and had no spontaneous respiration. Positive pressure ventilation with bag and mask was performed and resulted in a partial improvement of cyanosis. Numerous endotracheal intubation attempts were unsuccessful, primarily because the endotracheal tube could not be passed through the larynx. Only ventilation through the tube inserted into the esophagus resulted in audible breath sounds over both lungs and improvement in oxygen saturation. The infant was transferred to the Neonatal Intensive Care Unit. The physical examination did not reveal any other abnormalities, but laryngoscopy confirmed that the endotracheal tube was in the esophagus with a normal supraglottic area and vocal cords. An attempt to intubate the endotracheal tube in the trachea was unsuccessful. Oxygenation was restored after re-inserting the endotracheal tube into the esophagus. The authors postulated that the patient had a tracheoesophageal fistula in addition to a subglottic obstruction, so ordered an emergency CT scan. The scan showed obliteration of the airway just below the level of the larynx. The orogastric tube and endotracheal tube were both placed in the esophagus (Fig. 1). At the carina, there was a small fistula connecting between the esophagus and the right proximal bronchus (Fig. 2) with hyperaeration in both lungs.

Emergency thoracotomy was performed. Operative findings revealed tracheal agenesis with a small fistula connecting the esophagus to the carina. A gastrostomy and thoracic esophageal ligation and esophageal intubation through cervical esophagostomy were performed to maximize ventilation through the fistula: notwithstanding, the child died at 5 days due to severe respiratory acidosis and hypoxia. Postmortem findings confirmed the absence of the trachea with a small fistula between the esophagus and carina.

DISCUSSION

In 1900, Payne identified tracheal agenesis in a cyanotic infant in whom the trachea could not

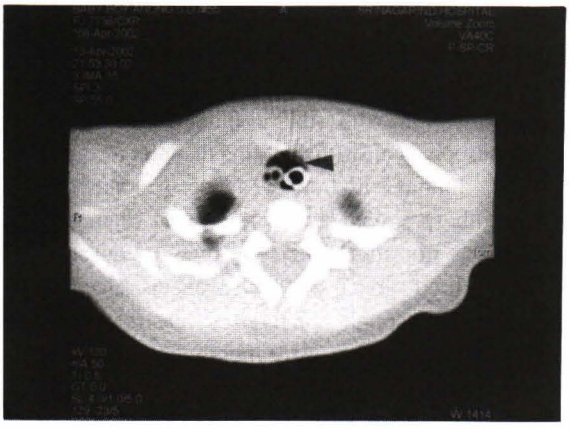

Fig. 1. Chest CT scan at the level of the thoracic inlet demonstrates the esophagus (arrow) containing both a double lumen orogastric tube and an endotracheal tube. The trachea is absent.

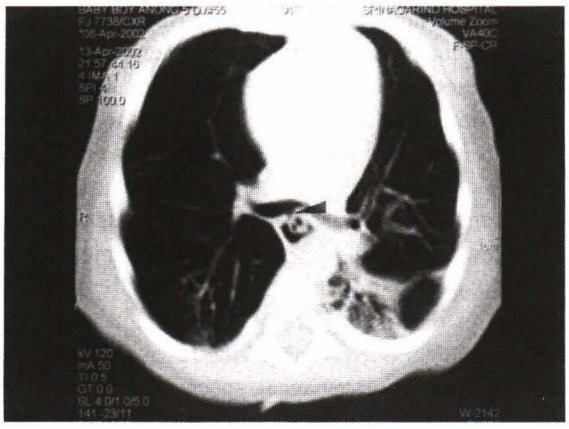

Fig. 2. Chest CT scan reveals a small fistula (arrow) between posterior aspect of the right proximal bronchus and the esophagus.

be found during an attempted tracheostomy[1]. Subsequent case reports of similar types of tracheal agenesis prompted Floyd[2] to develop a three-stage anatomical classification system: Type 1 refers to an agenesis of the proximal trachea with a normal, but short distal tracheal segment and a tracheo-esophageal fistula; Type 2 is an agenesis of the entire trachea with normal main bronchi communicating *via* the tracheo-esophageal fistula at the carina; and, Type 3 is a complete agenesis of the trachea and the right and left main bronchi arising separately from the esophagus. The presented patient had Floyd type 2, which is the most common form of agenesis.

Agenesis of the trachea occurs during embryonic development. One theory hypothesizes the lung buds occur before the foregut is divided into the esophagus and the trachea[3]. According to Sadler [4] the foregut forms two longitudinal ridges, which fuse to form a septum during the 4th week of gestation dividing the foregut into the esophagus dorsally and the trachea ventrally. Some of the earliest explanations of tracheal and esophageal malformations hypothesize either failure of displacement of the septum and/or the formation of ridges[5] or displacement of the dividing septum[6] with the degree of displacement determining the severity of the tracheal agenesis.

Alternatively, in a study of the foregut region of chick embryos, Kluth et al[7] observed that differentiation of the foregut into the esophagus and trachea was not a result of lateral ridges but a process of reduction in size of a foregut region called the tracheo-esophageal space. This reduction was caused by a system of folds that developed into the primitive foregut. They concluded that malformations of the trachea or esophagus with fistula could be explained by abnormalities in the formation of these folds or their developmental movements. For example, atresia of the trachea with fistula can be caused by a deformation of the foregut on the ventral side when the developmental movements of the folds are disturbed and the tracheo-esophageal space is dislocated in the dorsal direction. It, therefore, differentiates into the esophagus. Another possibility is that abnormal cellular interaction between the epithelium and mesenchyme leads to a disruption of the normal separation of the esophagus and trachea[8,9].

Tracheal agenesis is often associated with other congenital anomalies. Numerous cardiac defects have been reported, including valvular malformation, great vessels abnormalities, septal defects and cardiac hypoplasia[10-14]. Laryngeal abnormalities (i.e. elliptical cricoid cartilages) and vertebral defects (i.e. hemivertebra and fused vertebrae) have been described as have gastrointestinal tract abnormalities such as duodenal atresia and imperforate anus. Renal malformations and other genitourinary tract abnormalities have also been correlated. Orthopedic abnormalities range from hypoplastic thumb to absent radii. It has been suggested that tracheal agenesis is associated with VATER or VACTERL association, which includes vertebral defects, imperforate anus, tracheo-esophageal fistula, radial and renal dysplasia, cardiac and limb malformations[10,12,13,15-19]. There was no associated anomaly in the presented case.

Pregnancy may be complicated with polyhydramnios, although some are uneventful as was the case the authors have described. Infants with tracheal agenesis are born with severe respiratory distress and are unable to cry. Endotracheal intubation attempts are unsuccessful, laryngoscopy may reveal the blind pouch below the vocal cords.

Initial stabilization of affected infants requires the establishment of a secure airway and providing adequate ventilation *via* intubation of the esophagus. Since this procedure causes gastric distention, repeated gastric decompression will be necessary until distal esophageal banding can be done. The anatomy of the distal airway is usually visualized by a barium esophagogram as the barium passes through the tracheoesophageal fistula. Bronchoscopy or esophagoscopy may reveal the blind pouch or proximal communication with the esophagus but the distal tracheoesophageal fistula is often difficult to visualize.

Infants with tracheal agenesis in most reports were stillborn or died shortly after birth when surgical repair was not successful. Hiyama[20] described a case of Floyd type 2 tracheal agenesis successfully managed by esophageal ligation, gastrostomy, and esophageal intubation. At 9 months of age, a long T-tube was placed in the esophagus, the infant could breathe room air and later learned to speak by closing the orifice of the T-tube. At age three, the patient received a colonic graft to reconstruct the esophagus, after which he could eat in small amounts.

Several methods for tracheal replacement have been proposed. These include the use of allografts[21-23] prosthetic materials[24,25], autologous

tissue[26-28] or a combination of these materials[29, 30] but no satisfactory approach has yet been developed.

In summary, tracheal agenesis is rare and usually fatal. Early recognition and proper airway stabilization by esophageal intubation may allow short-term respiratory support. Attempts at surgical correction at the current state of technology are debatable, so parents should be counseled about the prognosis allowing them to make an informed decision. Increasing the knowledge and advances in surgical reconstruction may improve survival of some tracheal agenesis infants.

ACKNOWLEDGEMENTS

The authors wish to thank Mr. Bryan Roderick Hamman for assistance with the English-language presentation of the manuscript.

(Received for publication on January 15, 2003)

REFERENCES

1. Payne W. Congenital absence of the trachea. Brooklyn Med J 1900; 14: 568.
2. Floyed J, Cambell D, Dominy D. Agenesis of trachea. Am Rev Respir Dis 1962; 86: 557.
3. Sadler TW. Respiratory system. In: Sadder TW ed. Langman's Medical Embryology. 8th ed. Philadelphia: Williams and Wilkins; 1995: 260-9.
4. Merei J, Hasthorpe S, Farmer P, Hutson JM. Relationship between esophageal atresia with tracheoesophgeal fistula and vertebral anomalies in mammalian embryos. J Pediatr Surg 1994; 33: 58-63.
5. Keith A, Spicer J. Three cases of malformation of tracheoesophageal septum. J Anat Physiol 1906; 41: 1952.
6. Bremer J. In: Congenital Anomalies of the Viscera: Their Embryologic Basis. Cambridge, MA: Havard University Press; 1957: 27-31
7. Kluth D, Steding G, Seidl W. The embryology of foregut malformations. J Pediatr Surg 1987; 22: 389-93.
8. Calson BM. Digestive and respiratory systems and body cavities. In: Calson BM, ed. Human Embryology and Developmental Biology. 2nd ed. St. Louis: Mosby; 1999: 342-9.
9. Evans JA, Greenberg CR, Erdile L. Tracheal agenesis revisited: Analysis of associated anomalies. Am J Med Genet 1999; 82: 415-22.
10. Anderson C, Ahmed G, Cadwell C. Tracheal atresia associated with congenital absence of the radii. Cli Pediatr 1981; 20: 478-80.
11. Effmann EL, Spackman TJ, Berdon WE, Kuhn JP, Leonidas JC. Tracheal agenesis. Am J Roentgenol Radium Ther Nucl Med 1975; 125: 767-81.
12. Reilly JS, Adkins JC, Supance JS, Stool SE. Tracheal agenesis: A case report and literature review. Trans PA Acad Ophthalmol Otolaryngol 1984; 37: 55-9.
13. Milstein JM, Lau M, Bickers RG. Tracheal agenesis in an infant with VATER association. Am J Dis Child 1985; 139: 77-80.
14. Diaz EM Jr, Adams AM, Hawkins HK, Smith RJ. Tracheal agenesis: A case report and literature review. Arch Otolaryngol Head Neck Surg 1989; 115: 741-5.
15. Holinger LD, Volk MS, Tucker G Jr. Congenital laryngeal anomalies associated with tracheal agenesis. Ann Otol Rhinol Laryngol 1987; 96: 505-8.
16. Bradenkamp JK, Maceri Dr, Crockett DM. Tracheal agenesis: The importance of an endoscopic diagnosis. Otolaryngol Head Neck Surg 1989; 101: 688-92.
17. Quan L, Smith DW. The VATER association. Vertebral defects, anal atresia, T-E fistula with esophageal atresia, radial and renal dysplasia: A spectrum of associated defects. J Pediatr 1973; 82: 104-7.
18. Koltai PJ, Quiney R. Tracheal agenesis. Ann Otol Rhinol Laryngol 1992; 101: 560-6.
19. Khoury MJ, Cordero JF, Greenberg F, James JM, Erickson JD. A population study of VACTERL association : Evidence for its etiologic heterogeneity. Pediatrics 1983; 71: 815-20.
20. Hiyama E, Yokoyama T, Ichikawa T, Matsuura Y. Surgical management of tracheal agenesis. J Thorac Cardiovas Surg 1994; 108: 830-3.
21. Khali-Marzouk JF. Allograft replacement of the trachea. Experimental synchronous revascularization of composite thyrotracheal transplant. J Thorac Cardiovas Surg 1993; 105: 242-6.
22. Moriyana S, Shimiru N, Teramoto S. Experimental tracheal allotransplantation using omentopexie. Transpl Proc 1989; 21: 2596-600.
23. Sabas AA, Uez JB, Rojas O. Replacement of the trachea with dura mater. Experimental work. J Thorac Cardiovas Surg 1977; 74: 761-5.

24. Jacobs JR. Investigation into tracheal prosthetic reconstruction. Laryngoscope 1988; 98: 1239-45.
25. Shaha A, Dimaio T, Money S. Prosthetic reconstruction of the trachea. Am J Surg 1988; 156: 306-9.
26. Cohen RC, Filler RM, Konuma K. The successful reconstruction of thoracic tracheal defects with free periosteal grafts. J Pediatr Surg 1985; 20: 852-8.
27. Kon M, Van den Hoff A. Cartilage tube formation by perichondrium: A new concept for tracheal reconstruction. Plast Reconstr Surg 1983; 72: 791-7.
28. Letang E, Sanchez-Lloret J, Gimferrer JM. Experimental reconstruction of the canine trachea with a free revascularized small bowel graft. Ann Thorac Surg 1990; 49: 955-8.
29. Har-El G, Krespi YP, Goldsher M. The combined use of muscle flaps and alloplastics for tracheal reconstruction. Arch Otolaryngol Head Neck Surg 1989; 115: 1310-3.
30. Hayashi A. An experimental study of successful tracheal replacement using pedicled latissimus dorsi muscle flap and soft Marlex mesh. J Jpn Assoc Thorac Surg 1991; 39: 144-53.

ภาวะไม่มีหลอดลมแต่กำเนิด : รายงานผู้ป่วย

ผกาพรรณ เกียรติชูสกุล, พบ, จรรยา จิรประดิษฐา, พบ*, สุกัญญา ทักษพันธุ์, พบ*, จามรี ธีรตกุลพิศาล, พบ*, เบญจพร นิตินาวาการ, พบ**, เชิดชัย ตันติศิรินทร์, พบ****

ภาวะไม่มีหลอดลมเป็นความพิการแต่กำเนิดที่พบได้น้อยมากและผู้ป่วยส่วนใหญ่มักเสียชีวิต มากกว่าร้อยละ 90 ของผู้ป่วยจะมีความผิดปกติอื่น ๆ ร่วมด้วย โดยเฉพาะระบบหัวใจและหลอดเลือด ระบบทางเดินอาหาร ระบบทางเดินปัสสาวะ และระบบสืบพันธุ์ ทารกส่วนใหญ่มักจะมีอาการหายใจลำบากอย่างรุนแรง ร้องไม่มีเสียง และการใส่ท่อลมคอทำได้ยาก หรือไม่สามารถทำได้ เป็นผลให้เกิดภาวะขาดออกซิเจน รายงานทารกไม่มีหลอดลมแต่กำเนิด 1 รายที่มาด้วยปัญหาการหายใจล้มเหลวตั้งแต่แรกเกิด

คำสำคัญ : ภาวะไม่มีหลอดลม, ความพิการแต่กำเนิด

ผกาพรรณ เกียรติชูสกุล, จรรยา จิรประดิษฐา, สุกัญญา ทักษพันธุ์, จามรี ธีรตกุลพิศาล, เบญจพร นิตินาวาการ, เชิดชัย ตันติศิรินทร์
จดหมายเหตุทางแพทย์ ฯ 2546; 86: 1086–1090

* ภาควิชากุมารเวชศาสตร์,
** ภาควิชารังสีวิทยา,
*** ภาควิชาศัลยศาสตร์, คณะแพทยศาสตร์ มหาวิทยาลัยขอนแก่น, ขอนแก่น 40002