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CASE REPORT

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Identical twins concordant for pulmonary sequestration communicating with the esophagus and discordant for the VACTERL association

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Abstract Communicating bronchopulmonary foregut malformations (CBPFMs) are unusual congenital structures composed of a segment of lung tissue connected to the foregut. We present what we believe is the first reported case of identical twins concordant for CBPFM who are discordant for the VACTERL association. Their nonfunctional lung tissue was successfully removed and the fistulae were corrected, and they are expected to live normal life spans. We review the literature concerning these malformations and the proposed theories of their etiology. This case report of concordance in identical twins suggests that a possible genetic component to CBPFMs cannot be ruled out. The discordance for the VACTERL association implies that the etiology is most likely multifactorial.

Keywords Communicating bronchopulmonary foregut malformations · Pulmonary sequestration · VACTERL

Introduction

Communicating bronchopulmonary foregut malformations (CBPFMs) are unusual congenital structures composed of a segment of lung tissue connected to the foregut [1]. The etiology of these malformations has yet to be determined. Furthermore, the presence of familial predisposition has not been definitively documented. While there have been isolated reports of identical twins concordant for tracheoesophageal fistula [2, 3], this appears to be the first reported case of identical twins concordant for CBPFM who are, in fact, discordant for the VACTERL association.

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Case report

A set of monochorionic, monoamniotic 32-week twins was born by cesarean section on July 30, 2003, secondary to increasing, repetitive, variable decelerations. The pregnancy was complicated by mild polyhydramnios, and twin A was thought to have a possible congenital cystic adenomatoid malformation on prenatal ultrasound. Twin A, a 2,080-g female, was noted at birth to have mild respiratory distress and drooling, along with episodic choking and coughing. Chest radiographs shortly after birth (Fig. 1) demonstrated an air-filled proximal esophageal pouch and copious air in the stomach, findings consistent with a proximal esophageal atresia and distal tracheoesophageal fistula. At the time, both lungs appeared normal and were aerated. Twin A was also noted to have multiple vertebral and rib anomalies consistent with the VACTERL association. An echocardiogram demonstrated dextrocardia, a patent foramen ovale, and a large patent ductus arteriosus with a left-to-right shunt. A pouchogram demonstrated no proximal fistula (Fig. 2). The infant had good renal function and continued to have improved respiratory stability on day 1. No karyotypic abnormalities were demonstrated.

On day 2 of life, twin A underwent right posterior lateral thoracotomy, which confirmed the diagnosis of a proximal esophageal atresia with a distal tracheoesophageal fistula (Gross type C). The infant underwent fistula repair and esophagoesophagostomy. Chest radiographs on the initial postoperative days (Fig. 3a) demonstrated a hazy left hemithorax and complete opacification of the right hemithorax as well as a right-shifted cardiomediastinal silhouette. Subsequent chest radiographs (Fig. 3b) demonstrated near total improvement of left lung edema and atelectasis but continued opacification of the right lung. During this time, the infant was treated for presumed pneumonia and pulmonary edema with antibiotics and Lasix. She was successfully extubated on day of life 8. She continued to have mild respiratory distress following extubation.

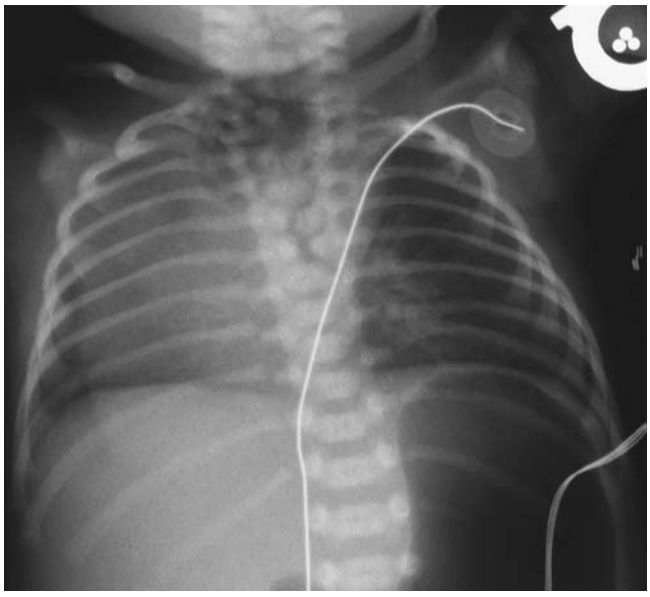

Fig. 1 Twin A, day 1: preoperative air-filled esophageal pouch and stomach, dextrocardia, and upper and midthoracic vertebral segmentation anomalies; both lungs aerated

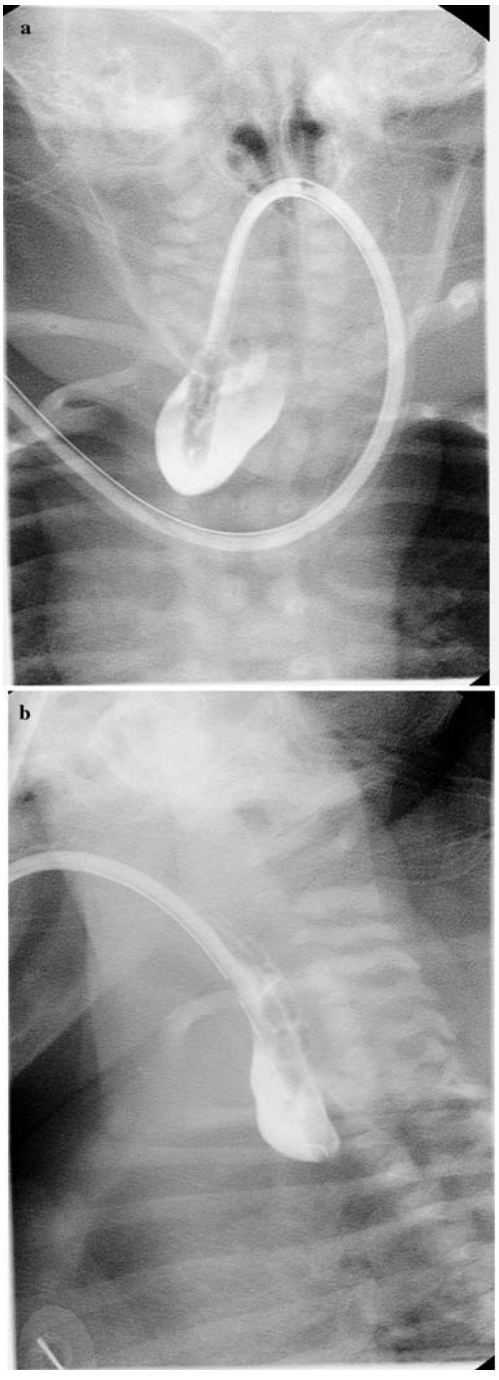

Fig. 2 Twin A, pouchogram, day 1. **a** AP. **b** Lateral

As a result, an esophagram was performed on day of life 9 (Fig. 4), which demonstrated a CBPFM from the right distal esophagus to the right lung. Subsequently, a thoracic computed tomography (CT) scan demonstrated a collapsed, contrast-filled lung located posterobasally in the right hemithorax and projecting superiorly to the level of the aortic arch. A complete absence of normal aerated lung tissue in the right hemithorax was demonstrated. The trachea deviated leftward at the expected location of the carina, and the right mainstem bronchus was absent.

On day of life 13, twin A underwent a repeat posterior lateral thoracotomy (transpleural) with resection of the right extrapulmonary sequestration and ligation of the CBPFM. The fistula was composed of esophageal tissue. The arterial supply to the sequestration was from a single pulmonary artery, and venous drainage was via two pulmonary veins. The sequestered lung appeared to have a hypoplastic bronchus connected to the esophagus. Postoperatively, twin A was extubated and had an uncomplicated recovery.

Twin B was a 1,950-g girl who initially required blow-by oxygen at birth to stimulate normal respirations. At the time, she was stable and able to breathe comfortably with oxygen by nasal cannula. Physical exam revealed coarse breath sounds on the right side. On day of life 1, the chest radiograph (Fig. 5a) demonstrated opacification of the left lung consistent with collapse or absence. A chest radiograph repeated on day of life 2 (Fig. 5b) showed continued opacification of the left chest. A CT scan performed on day of life 2 (Fig. 6) demonstrated a hypoplastic left lung, extrapulmonary sequestration, left bronchial atresia, and a small airway in the hypoplastic lung that appeared to connect with the esophagus. The arterial supply to the left sequestered lobe appeared to originate from branches of the thoracic aorta. An esophagogram demonstrated a bronchial connection with the distal esophagus (Fig. 7). Twin B subsequently underwent a left posterior lateral thoracotomy, revealing the absence of a left lung and the presence of a small extrapulmonary sequestration communicating with the esophagus through an esophageal fistula. The connection to the sequestered lung was consistent with esophageal tissue. The sequestration was supplied by two

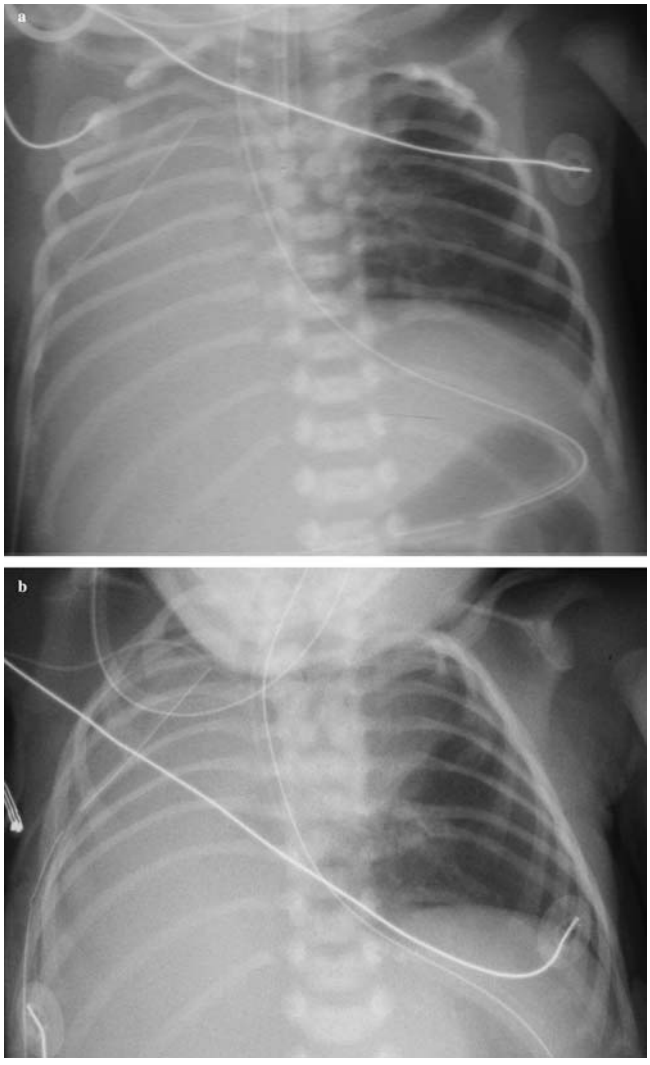

Fig. 3 **a** Twin A immediately postoperatively: nonaerated right lung. **b** Twin A, postoperative day 7: continued nonaerated right lung

individual branches of the thoracic aorta and was drained by a solitary pulmonary vein. The infant underwent excision of the left pulmonary sequestration with ligation of the feeding and draining vasculature and closure of the esophageal fistula.

Following her surgery, twin B was successfully weaned from the ventilator and was extubated on postoperative day 6. Her recovery was complicated by a chylothorax that appeared on postoperative day 15 and has since resolved.

Discussion

The term ''bronchopulmonary foregut malformations'' was introduced by Gerle et al. [4] to describe a spectrum of congenital malformations including intralobar and extralobar pulmonary sequestrations with or without

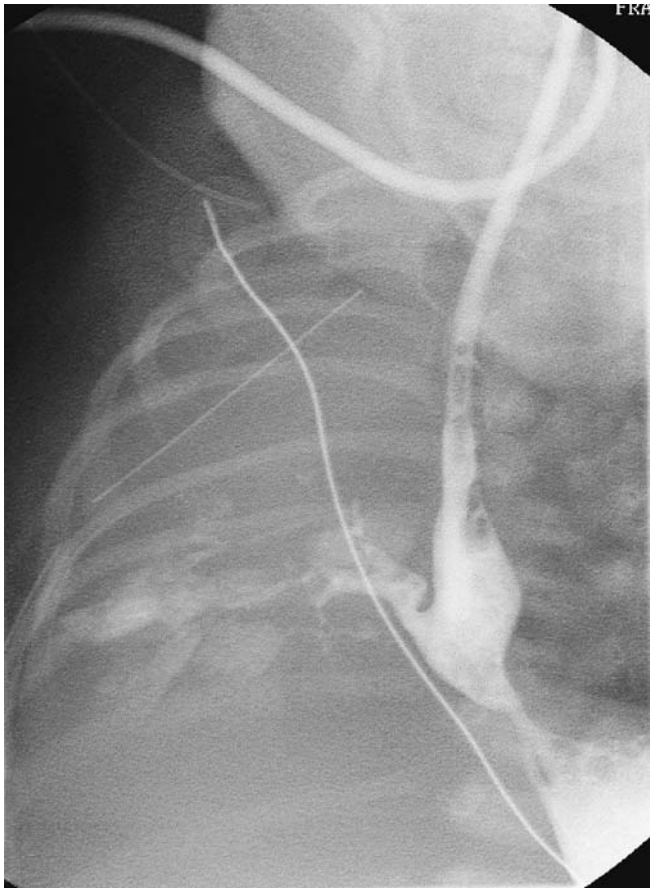

Fig. 4 Twin A, AP esophagogram, postoperative day 7: right bronchus arising from distal esophagus

patent connections to the alimentary tract. This expression has since been widely used to encompass a variety of malformations, including foregut duplication cysts, esophageal diverticuli, and cystic adenomatoid malformations [1, 5]. Lesions have been described in all lobes of the lungs with various blood supplies and in conjunction with several other congenital anomalies [1, 4, 5–15]. In addition, there is a spectrum of laryngotracheal abnormalities that may be associated with tracheoesophageal malformations as well as bronchopulmonary aberrations [16, 17].

The refined term ''CBPFM'' was suggested by Srikanth et al. [1] to specifically identify the very rare case of lung or lobar sequestrations with a patent connection to the esophagus or stomach. In addition, Srikanth et al. proposed an anatomical classification system for CBPFMs (Table 1). In our case report, both twins A and B typify this classification scheme. Twin A is an example of a group IA malformation—total sequestered lung associated with tracheoesophageal fistula and esophageal atresia [1]. Furthermore, she has other anomalies that indicate she is within the spectrum of the VACTERL syndrome. Whalen et al. noted that twins who have esophageal atresia and tracheoesophageal fistula are more likely to have complicated anatomy [18],

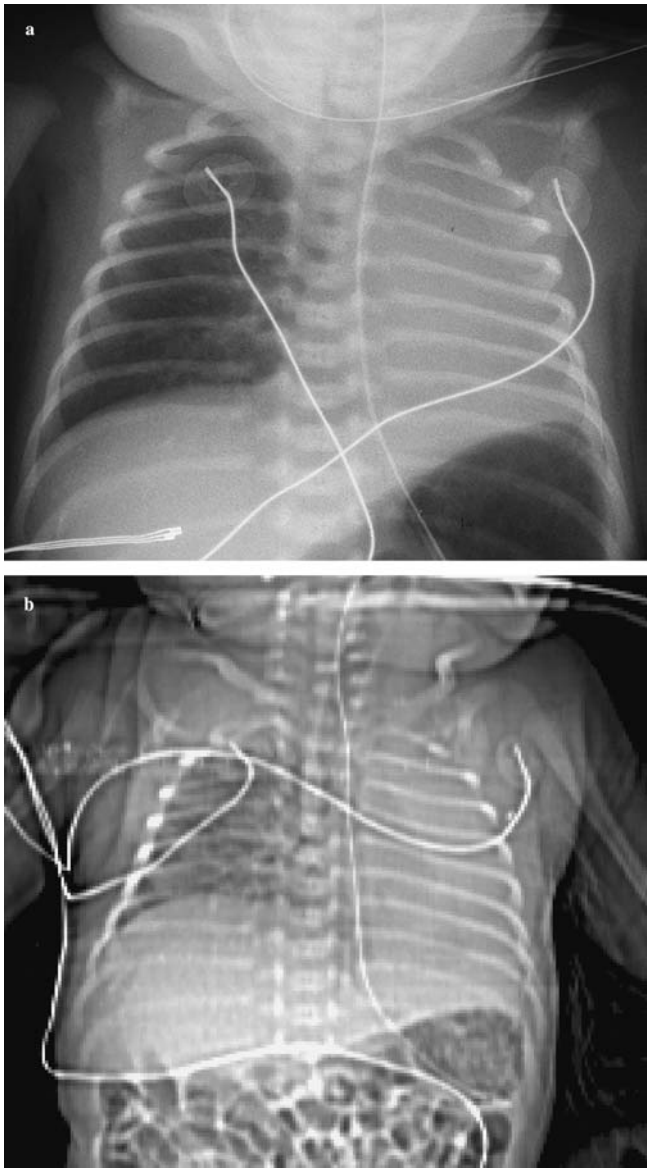

Fig. 5 **a** Twin B, day 1: collapsed vs. absent left lung. **b** Twin B, day 2: continued collapsed vs. absent left lung

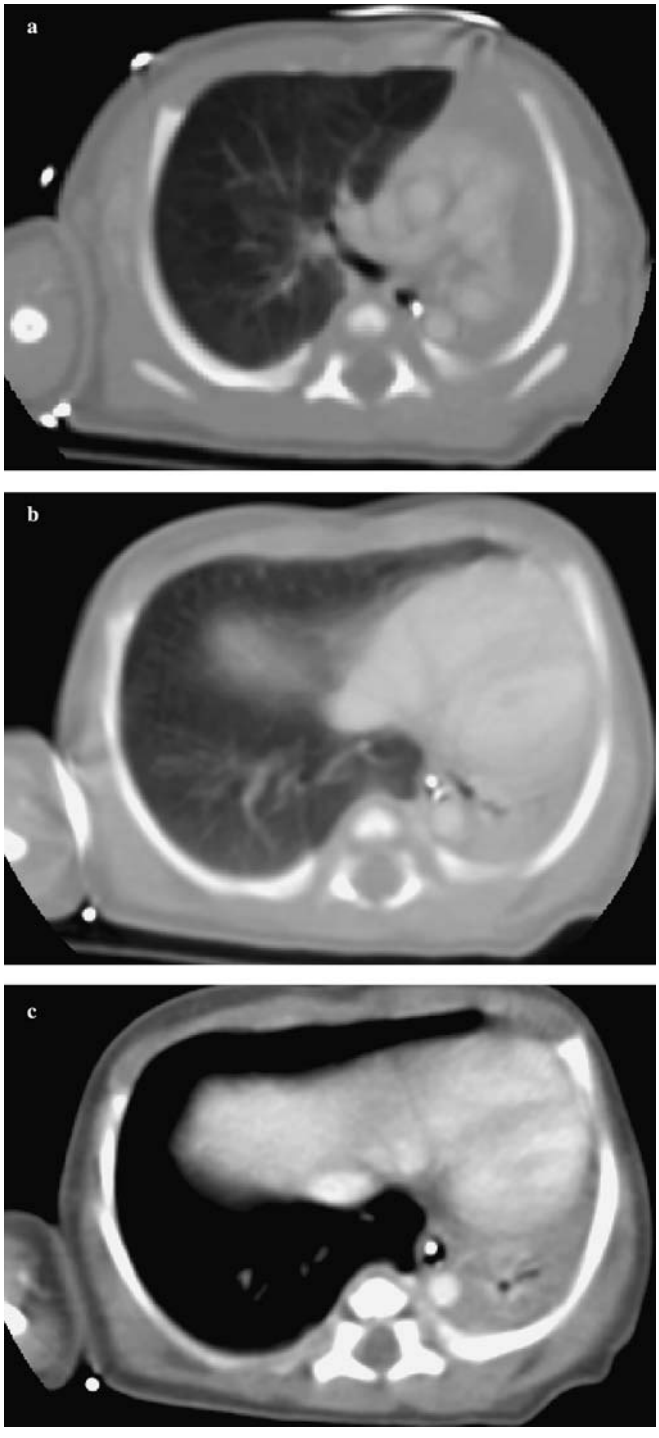

Fig. 6 Twin B, computed tomography, day 2. **a** Just below carina, right bronchus and esophagus with air and feeding tube; no left bronchus arising from the carina and no left pulmonary artery seen. **b** At base of thorax, tiny left bronchus arising from esophagus (feeding tube in esophagus). **c** Just above diaphragm, tiny left bronchus within small atelectatic left lung behind heart; tiny systemic artery extending from aorta to left lung; air and feeding tube within esophagus

including other malformations such as CBPFM. Twin B's malformation is within group II, which represented 33% of the CBPFMs in Srikanth et al.'s report [1]. Notably, 18 of 19 group II cases reported a sequestered right lung [1]; however, twin B had a left sequestration.

To date, no consensus has been formulated concerning the embryologic mechanism of CBPFMs. One theory postulates that these malformations are derived from an accessory or aberrant diverticulum of the esophagus composed of pluripotent cells that differentiate into pulmonary tissue [4, 5, 12, 19–21]. Srikanth et al. proposed that these malformations arise in days 32 through 41 of development as the lung buds migrate anteriorly to embrace the esophagus [1]. The stimulus or lack thereof for these malformations is unclear. This case report of concordance in identical twins suggests that a possible genetic component to CBPFMs cannot be ruled out. The discordance for the VACTERL association implies that the etiology is most likely multifactorial.

Table 1 Classification for communicating bronchopulmonary foregut malformations proposed by Srikanth et al. [1]

Group	Description
IA	Total sequestered lung communicating with the foregut, associated with esophageal atresia and tracheoesophageal fistula to the distal pouch
IB	Sequestered anatomic lobe or segment communicating with the foregut, associated with esophageal atresia and tracheoesophageal fistula to the distal pouch
II	Total sequestered lung communicating with the lower esophagus; absent ipsilateral mainstem bronchus
III	Isolated anatomic lobe or segment communicating with foregut
IV	Portion of normal bronchial system communicating with the esophagus

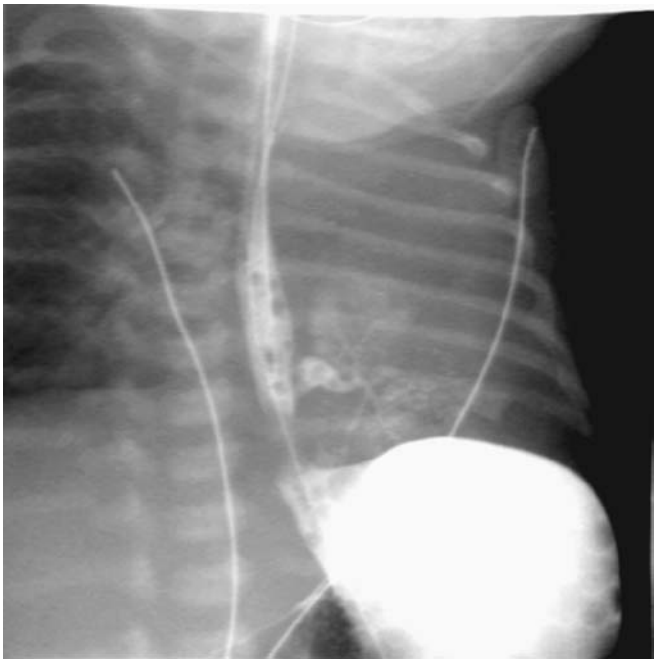

Fig. 7 Twin B, AP esophagogram, day 4: tiny left bronchus arising from distal esophagus, extending to small left lung

The significance of twinning in the production of CBPFMs and other associated malformations is not clearly defined. In general, concordance among twins is rare. Several reports of twins concordant for esophageal atresia with tracheoesophageal fistula have been described [2, 3]. Genetic predisposition and subsequent familial inheritance of tracheoesophageal fistula have been theorized [22], while other authors have indicated that the incidence of twinning is no greater than in the general population [18]. However, the VACTERL association is generally not associated with a familial history [22, 23].

A similar but often lethal combination of unilateral pulmonary agenesis combined with esophageal atresia and distal tracheoesophageal fistula has also been described [24–26]. This malformation has been linked to the VACTERL association. Unlike infants born with pulmonary agenesis, the infants in our case report had nonfunctional lung tissue that was successfully removed and fistulae that were able to be corrected, and they are expected to live normal life spans.

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