

Long-gap esophageal atresia associated with foregut duplication – a case report and literature review

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ABSTRACT

Coexisting long-gap esophageal atresia (EA) with a mediastinal duplication cyst is rare and difficult to diagnose. A 12-month-old boy, after multistage long-gap EA repair, presented with recurrent airway infections and a large mediastinal cystic lesion on computed tomography scans. Esophageal atresia repair was performed during infancy, with no coexisting pathology reported intraoperatively. Open Nissen fundoplication with gastrostomy and pyloroplasty was performed due to refractory gastroesophageal reflux. Recurrent airway infections were observed after

an asymptomatic period. Thoracoscopic removal of the foregut duplication cyst was performed. At the 8-month follow-up, no recurrence of symptoms was reported. The coexistence of a foregut duplication cyst and EA is very rare, and diagnosis is challenging. Only 14 cases have been reported in the available literature. We recommend considering the possible association of EA and congenital foregut cyst intraoperatively, especially in symptomatic patients.

Keywords: esophageal atresia; esophageal duplication; long gap; case report; foregut cyst.

INTRODUCTION

Esophageal atresia (EA) is the most common type of congenital disorder of the esophagus in newborns, with a prevalence of 1 in 4,000 live births [1]. The most commonly used classification of EA is the one proposed by Gross et al. in 1953 [2]. Esophageal atresia is often associated with the occurrence of a tracheoesophageal fistula (TEF). When the distance between the upper and lower pouch of the esophagus is too wide to perform primary reconstruction (gap length is more than 3 cm or more than 2 vertebral bodies), this condition is called long-gap EA [3]. Long-gap EA accounts for only 4–10% of all EA cases [4, 5].

Alimentary tract duplication occurs with a frequency of 1 : 4,500 live births and constitutes about 30% of foregut duplications. Esophageal atresia coexisting with other congenital anomalies is quite common, but cases of long-gap EA coexisting with a bronchogenic cyst or foregut duplication have been rarely reported in the literature [6, 7]. The rare occurrence and often asymptomatic course of foregut duplication make it very difficult to recognize, especially in the neonatal period. A typical preoperative diagnostic pathway in EA patients may not rule out coexisting pathology in the mediastinum.

We present a rare case of a young male patient with long-gap EA without a TEF (type A according to the Gross classification) and late manifestation of an esophageal duplication cyst. Clinical data, diagnostic evaluation, and details of surgical treatment, together with a current review of the literature, are presented. This manuscript was prepared following the case reports (CARE) guidelines [8].

CASE HISTORY

A 2,400 g male newborn, delivered by cesarean section at 37 weeks of gestation, was transferred to our department with a suspected congenital EA. Radiological examination confirmed EA without proximal or distal TEF (Fig. 1).

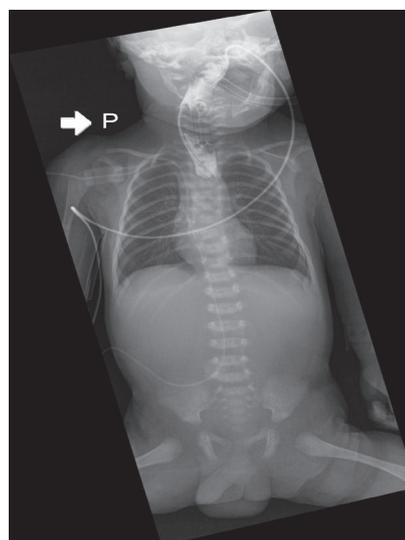


FIGURE 1. Preoperative radiograph study with contrast in the upper pouch of the atretic esophagus

Preoperative evaluation for coexisting anomalies revealed thoracic vertebral anomalies (Th8-9) and a small atrial septal defect. On the third day of life, a right-sided thoracoscopy was performed. The upper and lower esophageal pouches were dissected, and a TEF was excluded. The long-gap type was confirmed, and internal traction was applied.

The second stage of the operation was performed on the 12th day of life, but the distance between the esophageal segments was still too long to allow for a tension-free anastomosis. On the 24th day of life, an end-to-end esophageal anastomosis was performed over a feeding tube. During each thoracoscopic procedure, no other coexisting pathology was detected.

The postoperative course was complicated by cholestasis with mild elevation of liver transaminases. Conservative treatment with choleretics was successful.

Three weeks postoperatively, the patient developed feeding intolerance with significant saliva retention. Esophagography revealed a critical esophageal stricture at the anastomotic site, necessitating mechanical stricture dilatation with Savary–Gilliard dilators (Cook Medical, USA), which resulted in a good outcome.

Six weeks after esophageal reconstruction, the patient developed severe symptoms of gastroesophageal reflux (GER), which did not respond to conservative treatment (elevated upper body positioning, prokinetic drugs, proton pump inhibitors, and thickened milk formulas). Two episodes of apparent life-threatening events were observed. The patient was qualified for surgical treatment of GER, and a laparotomy with Nissen fundoplication was performed. The first stage of the procedure was initiated laparoscopically; however, due to insufficient working space secondary to intestinal distension, conversion to laparotomy was required.

Postoperative radiographs showed signs of esophageal dysmotility but excluded anastomotic stricture and refractory GER. Due to clinical signs of food aversion and feeding difficulties, a Kader–Stamm gastrostomy with concomitant Heineke–Mikulicz pyloroplasty was performed 1 month later. In the subsequent clinical course, the patient was fed via a gastrostomy tube with good tolerance.

According to our internal protocol, pulmonologic assessment was conducted due to signs of tracheomalacia. Bronchoscopy revealed mild segmental tracheomalacia in the lower third of the trachea, along with mild laryngomalacia.

Twelve months later, chest computed tomography was performed due to recurrent airway infections. A large mediastinal cystic lesion was identified in the middle mediastinum, near the hilum of the right lung, extending into the posterior mediastinum (Fig. 2).

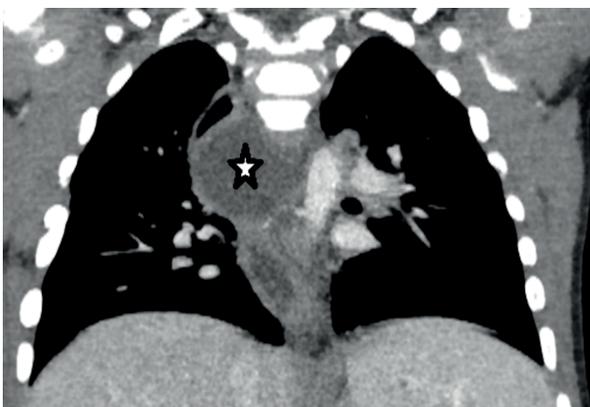


FIGURE 2. Computed tomography chest scan with a mediastinal mass (asterisk)

The patient subsequently underwent thoracoscopic resection of the mediastinal cyst via a right-sided approach, without opening the esophageal lumen, despite its close adherence to the esophageal wall in the upper part of the intrathoracic esophagus. Pathological examination confirmed a $2 \times 2 \times 1.5$ cm foregut duplication cyst lined with columnar epithelium and a double-layered wall.

The further clinical course was uneventful. Follow-up esophagography performed 3 months later excluded any anatomical abnormalities. The patient's oral intake improved, the diet was successfully expanded, and the removal of the gastrostomy tube is planned.

DISCUSSION

Esophageal duplication cysts are rare congenital anomalies, with an estimated incidence of 1 in 8,200 patients [9]. They likely result from a foregut budding error occurring between the third and sixth weeks of embryological development. Typically, the foregut divides into ventral and dorsal parts – the former gives rise to the laryngotracheal tree (respiratory tract), while the latter develops into the esophagus [10, 11].

The coexistence of a foregut cyst and EA, with or without TEF, is rare. Experimental studies have suggested that these 2 congenital disorders share a common origin, related to the failure of the foregut division process during embryonic development [12].

The diagnosis of an esophageal duplication cyst should be based on 3 characteristics:

1. a well-developed smooth muscle layer,
2. an epithelial lining representing a part of the alimentary tract (which may also include respiratory elements),
3. an attachment to the esophageal wall [13].

Foregut cysts can present in different morphological types: cystic, tubular, and diverticular. The most common type is cystic, which may communicate with the esophageal lumen or remain non-communicating. Tubular cysts are rare but more challenging to manage due to their typically long segment of the common wall with the esophagus and frequent communication with its lumen [14].

A histological feature of foregut duplication cysts is the presence of ciliated epithelium, which is found in both the alimentary and respiratory tracts [15]. Foregut cysts may be classified as: bronchogenic, enterogenous (including esophageal duplication), neuroenteric, or mixed [16, 17]. They are typically located in the posterior mediastinum [7].

Bratu et al. described 4 factors influencing the clinical presentation of this congenital anomaly: lesion localization, compression of surrounding structures (mass effect), complications due to glandular secretion, and infections [18]. The diagnosis of a foregut duplication cyst is often delayed and may occur at any age. Most of these lesions are asymptomatic and difficult to diagnose, but complications can arise. Cases described in the literature include complications such as: perforation, bleeding, rupture, infection, and malignant transformation [19, 20,

21]. In symptomatic cases, the most common presentation is respiratory symptoms due to the mass effect. The possibility of a mediastinal mass, particularly a foregut cyst, should be considered in newborns with respiratory obstruction [22, 23].

The coexistence of a foregut cyst and EA, with or without TEF, is extremely rare, with only 14 cases described in the literature according to the Gross classification. We have summarized the characteristics of these cases in a literature review (Tab. 1).

Nine of the 14 (64%) reported cases of foregut cysts associated with EA were diagnosed and excised in a 1-stage operation. The currently presented case is the first reported patient with long-gap EA coexisting with a duplication cyst. It is also the first case treated using minimally invasive techniques. In the authors' opinion, a thoracoscopic approach could be the preferred method, even in patients who have undergone multistage reconstruction of EA [24, 25].

Foregut cysts and EA carry a significant risk of complications, and surgical treatment is considered the standard of care [26, 27, 28]. Knod et al. reported that in cases of duplication cysts associated with EA, diagnosis is often delayed due to an asymptomatic presentation and the small size of the

lesion in the newborn period. It is rare for a foregut cyst to be visible during EA repair [29].

In our case, the cystic lesion could hypothetically have been diagnosed during the primary operation; however, such lesions are typically small and difficult to visualize in newborns. In cases where a duplication cyst is identified during EA (EA-TEF) repair, a 1-stage procedure is recommended. The goal of surgical treatment is to restore esophageal continuity while preserving the adjacent segment of the foregut cyst during resection, thereby reducing both short-term and long-term complication rates [30].

CONCLUSION

The coexistence of a foregut duplication cyst and EA is very rare, and diagnosis is challenging. We recommend considering the possible association of EA and congenital foregut cyst intraoperatively, especially in symptomatic patients presenting with dysphagia or obstructive airway symptoms during the preoperative course.

TABLE 1. General characteristics of reported cases of esophageal atresia (EA) associated with foregut duplication cyst in children

No.	Authors (year)	Type of EA (Gross classification)	Localization of duplication cyst	Primary (P) or delayed (D) diagnosis	Histopathology	Treatment	Coexisting anomalies
1.	KroczeK and Lipa (2025)	long-gap (type A)	posterior mediastinum; intramural duplication cyst, without communication to esophageal lumen	D	cystic lesion lined with columnar epithelium and 2-layer muscular wall	excision (thoracoscopy)	ASD II; vertebral anomaly Th8-9
2.	Spataru et al. (2015) [30]	type C	posterior mediastinum; intramural cyst of lower esophagus near TEF, without communication to esophageal lumen	P	cystic lesion lined with respiratory epithelium and 2-layer muscular wall	excision (thoracotomy)	none
3.	Knod et al. (2013) [29]	type C	posterior mediastinum; intramural foregut cyst of lower esophagus near TEF	P	cystic lesion lined with columnar and mucin-producing epithelium and 2-layer muscular wall	excision (thoracotomy)	trisomy 21
4.	Escobar Jr et al. (2012) [31]	type C	posterior mediastinum; large congenital diverticulum of esophagus	D	complex bronchopulmonary foregut malformation with ectopic pancreatic tissue	excision (thoracotomy)	vascular ring
5.	Inan et al. (2007) [32]	type C	posterior mediastinum; duplication cyst with communication to esophageal lumen	P	squamous epithelium with bundles of smooth muscle in the wall of the cyst	excision (thoracotomy)	none

TABLE 1. General characteristics of reported cases of esophageal atresia (EA) associated with foregut duplication cyst in children

No.	Authors (year)	Type of EA (Gross classification)	Localization of duplication cyst	Primary (P) or delayed (D) diagnosis	Histopathology	Treatment	Coexisting anomalies
6.	McNally et al. (2001) [10]	type C	thoracoabdominal, posterior mediastinum	D	mixed components: bronchogenic, esophageal duplication and pancreatic enterogenic cyst	excision (thoracotomy)	none
7.	Yamagiwa et al. (1998) [33]	type C	posterior mediastinum	P	small mass at distal end of proximal pouch of esophagus with pancreatic tissue	excision (thoracotomy)	none
8.	Snyder et al. (1996) [26]	type E (H-fistula)	posterior mediastinum; Y-shaped duplication of the esophagus	D	cystic duplication lined by squamous epithelium	excision (thoracotomy)	tracheomalacia
9.	Janik et al. (1988) [34]	type B	posterior mediastinum; esophageal duplication without communication to its lumen	P	cystic duplication with respiratory epithelium and mucous glands and 2-layer muscular wall	excision (thoracotomy)	cleft lip and palate
10.	Santamaria et al. (1988) [35]	type C	posterior mediastinum; cystic duplication	P	cystic duplication with columnar pseudostratified heterotopic epithelium	excision (thoracotomy)	none
11.	Narasimharao and Mitra (1987) [20]	type C	posterior mediastinum; cystic duplication near TEF	P	cystic duplication lined with pseudostratified columnar epithelium with smooth muscle bundles in the wall	excision (thoracotomy)	none
12.	Kirks and Filston (1981) [36]	type B	posterior mediastinum; cystic duplication attached to the membranous part of trachea, above carina	D	cystic duplication with stratified columnar epithelium and smooth muscle wall	excision (thoracotomy)	none
13.	Hemalatha et al. (1980) [37]	type C	thoracoabdominal; 1 cystic lesion in posterior mediastinum attached to lower right lobe; 1 tubular duplication along greater curvature of stomach	P	N/A; multiple duplications with pyloric-type gastric mucosa and pancreatic tissue with smooth muscle wall	stage excision (thoracotomy and laparotomy)	none
14.	Hemalatha et al. (1980) [37]	type A	posterior mediastinum; cystic duplication without communication to esophageal lumen	P	cystic duplication with squamous and transitional ciliated epithelium and 2-layer muscle wall	excision (thoracotomy)	none

ASD II – atrial septal defect type II; TEF – tracheoesophageal fistula

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