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DOI: 10.59736/IJP.23.01.919

CASE REPORT

A complex foregut malformation: case report and review

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ABSTRACT

Introduction: Esophageal duplication cysts (EDCs) associated with pulmonary sequestration (PS) are rare congenital anomalies originating from the embryonic foregut. They often present diagnostic challenges due to their complex nature and non-communicating features.

Case Report: We report the case of a one-year-old girl with a prenatal diagnosis of a cystic lesion, initially suspected to be congenital cystic adenomatoid malformation. Postnatal imaging confirmed the presence of PS and a non-communicating EDC. Thoracoscopic lobectomy was performed, and histopathology confirmed the diagnosis.

Discussion: This case highlights the difficulty in diagnosing non-communicating EDCs with PS, especially prenatally. Imaging techniques like CT and MRI are crucial, but histological analysis remains the gold standard for confirmation. Multidisciplinary management is essential for accurate diagnosis and effective treatment.

Conclusion: Early surgical intervention is critical to prevent complications and ensure a favorable outcome. Thoracoscopic surgery offers benefits in terms of morbidity reduction and preservation of lung function.

Keywords: Esophageal duplication cyst, pulmonary sequestration, foregut malformation.

This article may be cited as: Piro L, Gallo E, Roggero A, Stagnaro N, Vellone VG, Torre M. A complex foregut malformation: case report and review. Int J Pathol; 23(1):43-50. https://doi.org/10.59736/IJP.23.01.919

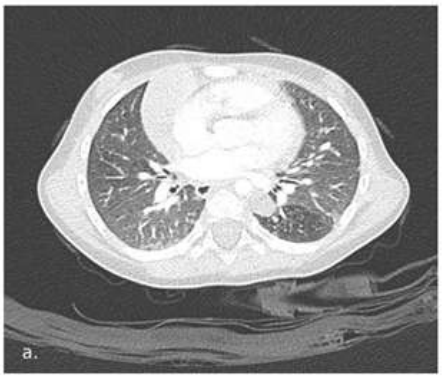
Introduction

The association of esophageal duplication cysts (EDCs) with congenital pulmonary malformations constitutes a complex and relatively rare phenomenon, involving the aberrant development of the embryonic foregut, which includes the bronchopulmonary respiratory pathways and the alimentary tract. EDCs represent a subset of these anomalies, originating from the foregut and causing symptoms and signs often related to the mass effect of an enlarged cyst. In children, common symptoms include stridor, dyspnea, wheezing, and dysphagia, while in neonates, frank respiratory distress is more likely. Imaging studies, such as chest computed tomography (CT) or magnetic resonance imaging (MRI), are crucial for evaluation, but diagnostic confirmation requires pathological examinations.[1,2] Pulmonary sequestration (PS) is a congenital lung anomaly described as a segment of lung not communicating with the normal bronchial system and receiving its blood supply from one or more anomalous systemic arteries. [3-7] PS usually presents as an isolated anomaly, but rarely it can be associated with other malformations, posing

a diagnostic and management challenge. [3,6] The coexistence of EDCs and PS is a very rare association [8–10], and generally, when present, they are communicating [11,12], configuring a picture of Bronchopulmonary Foregut Malformations (BPFM). We present a rare case of a non-communicating EDCs associated with a PS. The diagnostic complexities presented by such malformations often result in a delay or misdiagnosis, which further complicates the treatment planning. Additionally, the non-communicating nature of the esophageal duplication cyst in this case adds another layer of difficulty in achieving a definitive diagnosis early on.

Case Report

A 1-year-old girl with a prenatal diagnosis of a complex pulmonary anomaly was referred to our pediatric institution for surgical evaluation. The prenatal ultrasound at 24 weeks gestational age revealed an anechoic cystic lesion measuring 19x18 mm, with a suspected diagnosis of congenital cystic adenomatoid malformation (CAM) type II. A CT scan performed at 6 months after birth showed a cystic, round-shaped lesion in the apical segment of the left inferior lobe, close to the pulmonary hilum, associated with a pulmonary sequestration (PS) and a small tortuous afferent vessel originating from the distal thoracic aorta (Fig. 1).



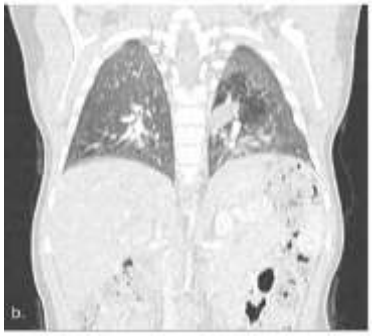


Fig.1 CT scan at 6 months of age. A. Axial scan. B. Coronal scan showing a cystic, round-shaped lesion in the apical segment of the left inferior lobe, close to an overaerated area of the lung parenchyma supplied by a systemic accessory vessel.

Six months later, an additional CT scan performed as part of the surgical planning showed that the malformation had remained stable. (Fig. 2)

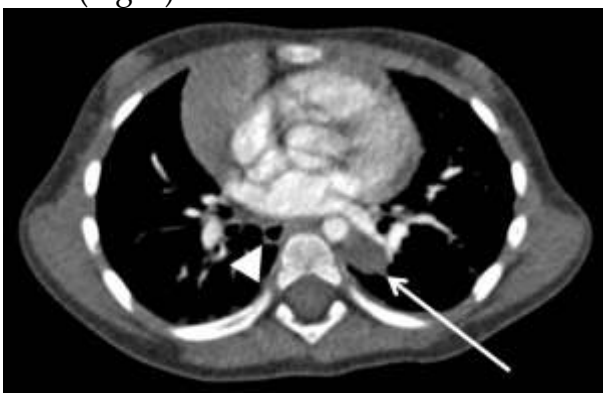


Fig. 2. Axial CT scan of the thorax after contrast medium injection reveals a hypodense oval lesion (arrow) adjacent to the descending aorta and the left lower pulmonary vein.

The differential diagnosis includes a pericardial cyst, bronchogenic cyst, and, less likely, CPAM (Congenital Pulmonary Airway Malformation), and pulmonary sequestration. Due to the distance from the esophagus (arrowhead), the hypothesis of esophageal duplication cyst has not been considered.

The patient underwent a thoracoscopic left lower lobectomy with the section of the afferent systemic vessel of the PS under

single pulmonary ventilation. A paravertebral analgesia catheter for post-operative pain control and a 12Ch chest drain were placed at the end of the procedure. The chest drain was removed on the second post-operative day, and the girl was discharged on the third post-operative day in excellent clinical condition.

Microscopic examination revealed lung tissue with dilated and non-aerated bronchi associated with an extremely thickened afferent artery. Signs of the systemic-pulmonary shunt, such as dilated vessels, were found in the subserous area of the visceral pleura. All characteristics were diagnostic for intralobar pulmonary sequestration (IPS). On macroscopic examination, a 9mm cyst was described close to the resection margin. The histologic examination of the cyst showed a multi-layered squamous epithelium p63+, CK7+, CK20-, with fetal-type squamous cells and ciliated columnar cells. Some components of tubulo-acinar glands were also found in the context of the sub-mucosa. The muscular tunica was irregular with some ganglia similar to those of the myo-enteric plexus. The histologic diagnosis provided evidence of IPS associated with an EDC without communication between the two anomalies. (Fig 3)

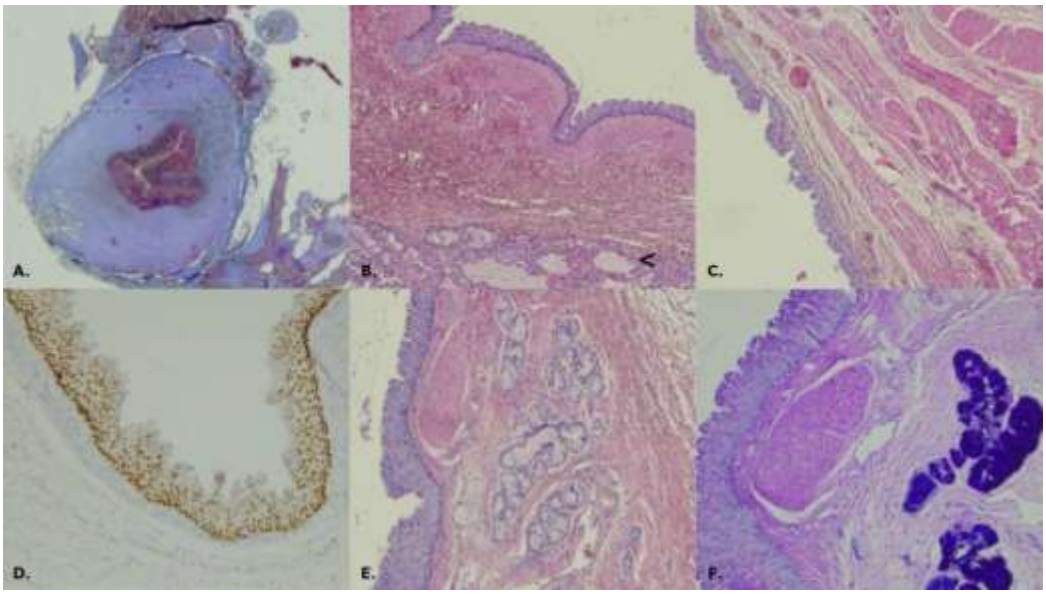


Fig 3. Figure 3: Microscopic examination of the specimens

A. H+E stain, 2.5x magnification, demonstrates a thickened-wall afferent artery of the IPS.
B. H+E stain, 5x magnification, shows the full-thickness cystic wall with both squamous and respiratory epithelium, some layers of fibromuscular tissue in the context of the sub-mucosa-associated smooth muscle layer, and an outline of a myenteric plexus-like structure. All features are consistent with an esophageal duplication cyst. The black arrow indicates pulmonary parenchyma filled with mucus, non-aeriated, typical for IPS.
C. H+E stain, 5x magnification, displays the full-thickness cyst wall with abundant muscular structures of the submucosa and a muscular layer below with associated ganglia-like structures.
D. Epithelium P63+, 10x magnification, details the presence of squamous epithelium lining the cyst wall consistent with an esophageal duplication cyst.
E. H+E stain, 5x magnification, provides a detail of the cyst's wall showing the presence of exocrine glands in the submucosal layer.
F. Alcian PAS Blue, 10x magnification, presents a detail on the exocrine glands of the cyst's wall.

At the six-month post-operative check, the patient was in excellent clinical condition, and the CT scan confirmed the resolution of the anomaly.

Discussion

The association of PS with esophageal duplication cysts (EDCs) represents a complex and relatively rare phenomenon. This condition poses significant challenges from both radiological and surgical perspectives. Generally PS presents as both isolated anomalies and as part of more complex BPFM. [3,4] The latter commonly involve foregut malformations in communication with cystic anomalies of the upper digestive tract, such as the EDC. [1,4,6] Our case represents a rare combination of pulmonary malformation associated with non-communicating EDC, which has never been described in the literature. Radiological evaluation is crucial for diagnosing and managing PS and EDCs. Prenatal imaging, such as ultrasound, often detects cystic lesions but may not reveal the full complexity of the malformations. This is a key strength of modern obstetric care; however, reliance solely on prenatal ultrasound can often be insufficient in complex cases. While ultrasound can identify cystic anomalies, its ability to reveal the full complexity of malformations like PS and EDC is limited. In the presented case, an anechoic cystic lesion was identified at 24 weeks gestation, initially suspected to be a congenital cystic adenomatoid malformation (CAM) type II. However, it was only through postnatal imaging that the complete picture emerged. Computed tomography (CT) scans, performed six months after birth, confirmed a cystic lesion in the left inferior lobe and revealed an associated PS with an anomalous systemic artery. CT scans, while excellent for delineating anatomical structures, may still struggle to accurately diagnose the full scope of foregut-related malformations, highlighting the need for complementary imaging techniques such as MRI, which offers superior soft tissue contrast. The use of magnetic resonance imaging (MRI) can provide additional insights into the anatomical relationships between these anomalies, offering better soft tissue contrast and aiding in the differential diagnosis of EDCs versus other cystic lung lesions. [1,12,13] In our case, prenatal diagnosis only revealed the IPS, while the association with EDC emerged only during postoperative analysis of the sample. In fact, due to the distance and lack of communication between the esophagus and the cyst, the diagnosis of esophageal duplication cyst had not been considered. In these complex malformations, radiological evaluation, including prenatal CT, often may underestimate the anomaly's complexity, highlighting the importance of clinical-radiological correlation. One notable limitation of the imaging studies in this case is the underestimation of the anomaly's complexity during prenatal imaging, which only revealed the pulmonary malformation without consideration of the potential esophageal duplication cyst. This emphasizes the need for a multimodal imaging approach, especially in cases where foregut malformations are suspected. Magnetic resonance imaging (MRI) could have provided additional information by offering better soft tissue contrast, which would have aided in differentiating EDCs from other cystic lung lesions like bronchogenic cysts or CPAM [13,14]. Accurate diagnosis requires pathological confirmation. Histological analysis of complex foregut anomalies is crucial for distinguishing between various types of malformations. In this case, the

histological examination of the cyst showed characteristics consistent with an EDC, including multi-layered squamous epithelium and ciliated columnar cells, which were essential for distinguishing it from other conditions such as bronchogenic cysts or congenital pulmonary airway malformations (CPAM). Given the frequent histological similarities between bronchogenic cysts and EDCs, diagnostic accuracy hinges on a detailed pathological examination, particularly in distinguishing between multilayered squamous epithelium and respiratory epithelium. This case is further complicated by the lack of communication between the PS and EDC, which is unusual and represents a unique diagnostic challenge.

Surgical intervention remains the primary treatment modality for both PS and EDCs, particularly to mitigate risks such as respiratory distress, infection, and potential malignant transformation. [3 4,8] Malignant transformation is a risk to be considered for both IPS and EDC, emphasizing the need for a thorough risk-benefit assessment prior to intervention.[2,9,12,15,19,25] Preoperative imaging plays a pivotal role in surgical planning, and in this case, serial CT scans were instrumental in assessing the stability of the malformation and planning the surgical approach. A multidisciplinary approach involving radiologists, pediatric surgeons, and pulmonologists is essential for managing such complex cases effectively.

Despite the lack of consensus on the timing of surgical intervention, an early approach is preferable to reduce the risk of complications. [1,6,9,12,14-24] The early definitive treatment reduces complication and conversion rates when performed through thoracoscopy, minimizing radio exposure during follow-up scans to monitor the lesion and allowing for better expansion and vicariant function of lung parenchyma. [12,13,23] This is beneficial, considering that the thoracotomy approach, which may be required due to complications' sequelae, is associated with musculoskeletal impairment and poorer cosmetic results. [23] Despite the proven safety and efficiency of elective thoracoscopic approaches in high-volume centers by expert thoracoscopic surgeons, the literature evidence remains limited.[26]

The choice between atypical lung resection and lobectomy should be tailored to the extent of the lesion and potential complications, with a focus on preserving as much lung parenchyma as possible to maintain lung function and minimize postoperative impairment.

Conclusion

Complex malformations like this pose a true diagnostic challenge and necessitate appropriate surgical intervention to prevent complications, including the potential for malignant transformation. In our opinion, an early thoracoscopic surgery offers significant advantages, including reduced morbidity and preservation of lung function. Despite the lack of consensus on the exact timing of surgical intervention, an early approach is generally preferred to reduce complications and optimize outcomes. Managing such cases requires a personalized and multidisciplinary approach involving radiologists, pulmonologists, and pediatric surgeons. In conclusion, managing non-communicating EDCs associated with PS requires a nuanced approach, integrating detailed radiological assessment with timely and appropriate surgical intervention.

For this article, no funding was received.
Conflicts of interest: The authors declare that they have no conflict of interest.

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HISTORY	
Date received:	10-10-2024
Date sent for review:	06-03-2025
Date received reviewers comments:	21-03-2025
Date received revised manuscript:	25-03-2025
Date accepted:	25-03-2025

CONTRIBUTION OF AUTHORS	
Contribution	**Authors**
Conception/Design	LP, EG, AR
Data acquisition, analysis and interpretation	NS, VV, MT
Manuscript writing and approval	LP, EG, MT

All the authors agree to take responsibility for every facet of the work, making sure that any concerns about its integrity or veracity are thoroughly examined and addressed.