

Esophageal duplication cyst presenting with stridor in a child with congenital pulmonary airway malformation

A case report and literature review

Ching-Fang Sun, MD^a, Chieh-Ho Chen, MD^b, Pin-Zuo Ke, MD^c, Tzu-Lung Ho, MD^d, Chien-Heng Lin, MD^{b,e,*}

Abstract

Rationale: Esophageal duplication cyst (EDC) is a rare developmental aberration originated from the embryonic foregut. It may remain asymptomatic but produce local mass effect on surrounding organs if rapid enlarges. EDC may sometimes accompany with other congenital malformations. Congenital pulmonary airway malformation (CPAM) is a congenital lung malformation with an unknown chance of developing symptoms. Here we report a rare case of esophageal duplication cyst with type 2 congenital pulmonary airway malformation (CPAM).

Patient concerns: A 16-month old boy with a prenatal diagnosis of type 2 CPAM presented progressive stridor and respiratory distress and was admitted to our hospital under the diagnosis of pneumonia. The patient responded poorly to antibiotics. A chest Xray (CXR) showed consolidation over the left upper lobe with trachea deviated to right side. A chest computed tomography (CT) revealed a cystic lesion sized 3.3 × 3.3 cm in the superior mediastinum.

Diagnoses: Post-operative pathological report confirmed the diagnosis of esophageal duplication cyst.

Interventions: We pre-medicated the patient with steroids and inhaled bronchodilators for airway maintenance. Then the patient received tumor resection via median sternotomy.

Outcomes: The patient recovered without complication and discharged smoothly 4 days after the surgery.

Lessons: EDC is a rare but potentially life-threatening disease owing to compression of large airways. Chest CT scan could detect the lesion non-invasively and should be considered in patients with persistent stridor, as well as CXR findings of the trachea deviated by a mass lesion in mediastinum, especially for those with CPAM.

Abbreviations: CPAM = congenital pulmonary airway malformation, CT = computed tomography, CXR = chest X-ray, EDC = esophageal duplication cyst, MRI = magnetic resonance imaging.

Keywords: congenital pulmonary airway malformation, esophageal duplication cyst, mediastinal cysts, pediatric

1. Introduction

Esophageal duplication cyst (EDC) belong to the spectrum of developmental aberrations arising from the embryonic foregut (bronchopulmonary and alimentary tracts). Symptoms and signs

often are resulted from a mass effect caused by an enlarged cyst. Stridor, dyspnea, wheezing, and dysphagia are common clinical features of EDC in children; while frank respiratory distress is more likely in neonates. Imaging study with chest computed tomography (CT) or magnetic resonance imaging (MRI) are valuable for evaluation, but confirmation with pathological findings are necessary to establish a diagnosis.^[1,2]

Congenital pulmonary airway malformations (CPAMs) are characterized by cystic and/or adenomatous lung areas. Nowadays, they are mainly antenatally diagnosed by sonography, but the natural evolution and chance of developing symptoms are hard to access. They may spontaneously resolve (11–49%), regress (18–42%), or progress (33–44%). Even while remaining asymptomatic, CPAMs still carry an increased risk of infections and malignancies.^[3] Only 3 cases of coexistence of both EDC and CPAM have been reported in the literature^[5–7] (Table 1). To the best of our knowledge, this is the first case report of EDC with left side type 2 CPAM. Our patient is also the only known case to recover well without lobectomy of CPAM.

2. Consent

Institutional Review Board (IRB) approval was not required for this case report. The patient's mother signed the necessary

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^a College of Medicine, China Medical University, ^b Division of Pediatric Pulmonology, China Medical University Children's Hospital, ^c Department of Pathology, China Medical University Hospital, China Medical University,

^d Department of Medical Imaging, China Medical University Hospital,

^e Department of Biomedical Imaging and Radiological Science, College of Medicine, China Medical University, Taichung, Taiwan.

* Correspondence: Chien-Heng Lin, Division of Pediatric Pulmonology, China Medical University Children's Hospital, No 2, Yuh-Der Road, Taichung 404, Taiwan (e-mail: lch227@ms39.hinet.net).

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Table 1
Summary of cases with coexistence of both esophageal duplication cyst (EDC) and congenital pulmonary airway malformation (CPAM).

References	Age, sex	Initial symptoms	EDC location/ CPAM location (type)	Surgical intervention	Complication
Kuga ^[5]	7 m, F	Productive cough and recurrent fever for 2 months	Right posterior mediastinum/ Right lower lobe (Type 1)	Cystectomy Right lower lobectomy	N/A
Hasegawa ^[6]	6 m, F	High fever and cough for 2 days	Right upper and middle lobes/ Right lower lobe (Type 1)	Cystectomy Right lower lobectomy	Pneumonia (<i>H. influenzae</i>)
Hani ^[7]	12 y, M	Repeated respiratory infection for 6 years	Posterior mediastinum/ Right upper lobe (Type 1)	Cystectomy Right upper lobectomy	Pneumonia without improvement with antituberculous
Our Case	1-y- and 4-m, M	Cough for 2 weeks and progressive stridor	Superior mediastinum/ Left upper lobe (Type 2)	Cystectomy	Pneumonia with tracheomalacia

F = Female, M = Male, m = month, N/A = not available, y = year.

documents to consent to the use of the patient's data for teaching and publication purposes.

3. Case report

A 16-month-old boy with a history of type 2 CPAM of the left lung was diagnosed at 21th week of gestational age and presented to our outpatient department because of cough, rhinorrhea, and post-tussive vomiting for 3 consecutive days (Fig. 1A). Physical examination was within normal limits except for coarse breathing sounds. The chest X-ray (CXR) showed consolidations over the left upper lobe. Despite initial treatment with antibiotic amoxicillin/clavulanic acid and azithromycin, his symptoms were exacerbated with hoarseness and stridor in the following 2 weeks. A repeated CXR 5 days after admission showed regression of pneumonia patch but also a mediastinal mass with obvious tracheal deviation to the right side (Fig. 1B). A chest sonography revealed a homogeneous hyperechoic cyst sized $3.6 \times 3.0 \times 2.4$ cm over the suprasternal area. A subsequent CT demonstrated a well-encapsulated cystic lesion up to 3.3×3.3 cm in size at the superior mediastinum, which contained a homogenous content and alveolar consolidation lesion in the left upper lung consistent with lung infection (Fig. 1C). A laboratory study with serum α -fetoprotein, serum β -human chorionic gonadotropin, and urine vanillylmandelic acid for tumor survey were all negative.

A bronchogenic cyst was initially suspected according to the chest CT. Thus, we suggested that the patient undergo surgical excision via median sternotomy. Considering possible inflammation related bronchial spasm and a mass compressive effect, we prescribed corticosteroids with methylprednisolone, bronchodilator with salbutamol, fenoterol, and aminophylline prior to operation. During the surgery, we found an isolated cystic lesion located between the trachea and bilateral carotid arteries. The cyst was completely removed and some yellowish turbid was exuded when the cyst was incised (Fig. 1D). Microscopically, the cyst was lined by squamous epithelium (90%) and ciliated columnar epithelium (10%). Also, there were some focal ulcerations with lymphoplasma cells, and neutrophil infiltration. The cyst wall presented predominantly of smooth muscles, with an absence of mucus glands, cartilages, thyroid follicles, lymphoid tissues, or thymic tissues. (Fig. 2) Finally, pathologic diagnosis was confirmed as an EDC.

The postoperative course was uneventful and smoothly, and he was discharged 4 days after surgery without any symptoms.

4. Discussion

The incidence of mediastinal lesions peaks in patients less than 10 year-olds (23.0%) with male predominance.^[8] Imaging with CT and MRI are important tools for the evaluation of mediastinal mass, but several types of lesions at superior mediastinum have similar characteristics in image. Duplication cysts (bronchogenic cysts, EDC, and neuroenteric cysts), dermoid cysts, and thyroglossal duct cysts can present as solitary, smooth, and thin-walled cysts containing non-enhancing, homogeneous, and attenuated fluids. In our case, bronchogenic cyst is impressed initially, as it was the most common foregut duplication cyst in young children. Dermoid cysts and thyroglossal duct cysts had once been considered, but were less preferred because merely 15% dermoid cysts contain cystic lesions without fat, nor any calcification; thyroglossal duct cysts are more likely to be found in the lateral infrahyoid neck.^[9,10]

Common symptoms of EDC include stridor, breathlessness, wheezing, and dysphagia. If ectopic gastric epithelium is presented, it may cause intra-cystic hemorrhage, ulceration, and fistulation. The result of fistulation into the tracheobronchial tree includes hemoptysis and pneumonia while hematemesis and melena may occur if the cyst is communicating with the esophagus.^[11] If symptoms at presentation concern malignancy, additional laboratory tests should be ordered to rule out tumors with high incidence in pediatric patients such as: α -FP and β -HCG for seminoma and non-seminomatous germ-cell tumors, lactate dehydrogenase (LDH) for lymphoblastic non-Hodgkin lymphoma, serum alkaline phosphatase (ALP), and white blood cell count for Hodgkin disease/mediastinal large cell lymphoma.^[11] Despite the above assessment, surgical specimen with histopathologic study is absolutely required for a definitive diagnosis.

Early surgical resection is recommended for both symptomatic and asymptomatic patients with EDC. Recurrent or persistent pulmonary infection are common complications of the developmental aberrations. Since increased rate of infection over time makes the surgery more difficult, early elective surgery is recommended.^[4] Carefully reviewed the literature, we found all cases of coexistence of both EDC and CPAM presented with the diagnosis of pneumonia or signs indicating infection such as fever (Table 1). Although the cyst may remain chronically asymptomatic, it may suddenly increase in size as a result of infection, intra-cystic hemorrhage, or tissue perforation from the presence of acid-secreting gastric epithelium. Even without trigger events, the cyst maybe slow-growing within its natural course. As a part of the endothelial lining, mucoid/serous

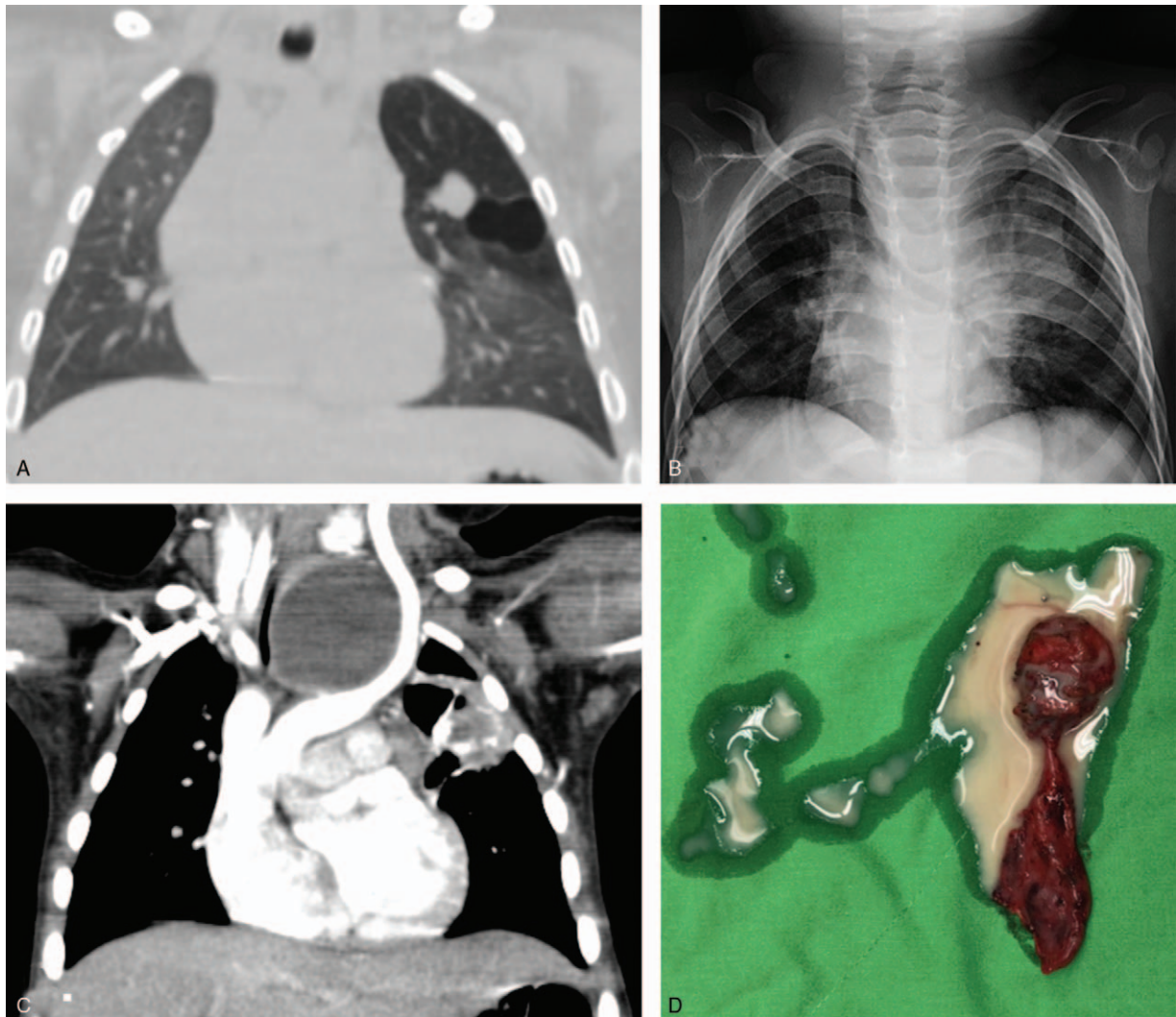


Figure 1. (A) The chest CT after birth showing a cystic lesion measuring 2.1×1.3 cm at left upper lung field with some solid component, consistent with CPAM. (B) The CXR showed consolidation in the left upper lobe and the trachea was deviated to the right side. (C) The CT showed a well-encapsulated cystic lesion up to 3.3×3.3 cm in the left superior mediastinum, which contained homogeneous content and alveolar consolidation lesion in the left upper lung consistent with lung infection. (D) The cyst contained some yellowish turbid fluid and there was no fistula, adhesion, nor focal inflammatory change.

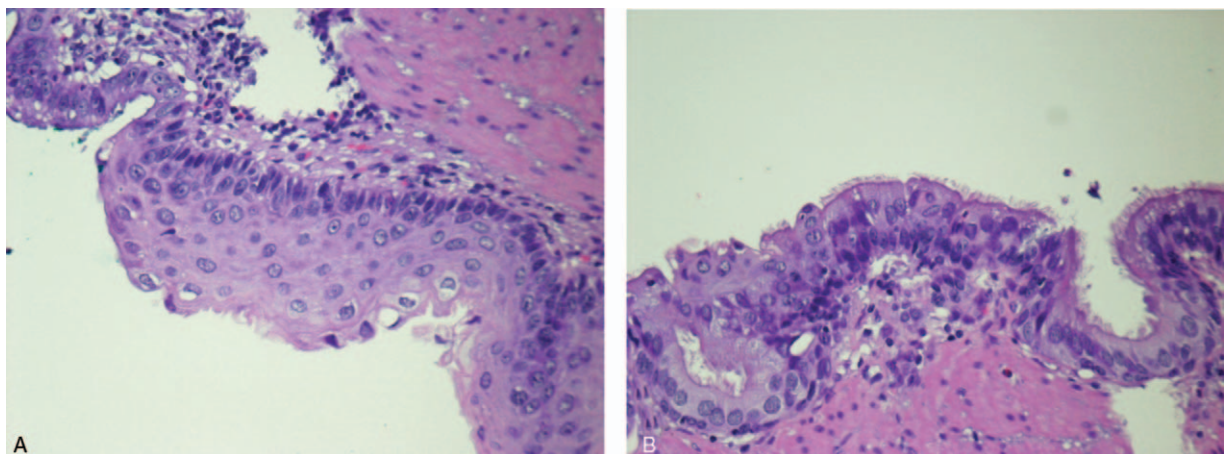


Figure 2. (A) The cystic epithelial lining presents predominantly stratified squamous epithelium. (B) A focal ciliated columnar epithelium is noted without associated gland or cartilage.

secreting cell directly secretes into the cyst while squamous epithelium shedding creates an intra-cystic osmolality that causes a fluid shift. Focal inflammatory changes induced by pneumonia may explain the sudden onset symptoms of our patient. The cyst increased in size rapidly in response to inflammation in the surrounding area. The pathologic report proved our case to be an example of uninfected cyst with a mere inflammatory change. Focal ulcerations, lymphoplasmic cell infiltration, and the absence of neutrophil and pathogens suggested inflammation instead of infection.

The histopathologic characters of EDC give us a hint of their common origin with CPAM. They are traditionally defined as cystic lesions that are covered by smooth muscle layer without cartilage. The lining of walls can be one or more types of epithelium, with an origin from either alimentary tract, respiratory tract, or both. The presence of ciliated columnar epithelium in our case indicated an origin from respiratory diverticulum, i.e., the early stage of pulmonary bud pouching out from the ventral part of the foregut.

The coexistence of EDC and CPAM is rare but not surprising, since they belong to the same spectrum of bronchopulmonary foregut malformations.^[12] Several embryological theories exist as the etiology of EDC:

1. development of an accessory diverticulum
2. duplication of epithelium that is supposed to obliterate during recanalization of gastrointestinal tract, and
3. a persistent endodermal-ectodermal tract that resulted in a cyst outside the vertebral column.^[11]

One theory for the development of CPAM is a genetic defect that interrupts lung morphogenesis when the trachea separates from the foregut. Another theory is the obstructive hypothesis, which states that a focal obstructive event engenders focal arrest in lung development at different stages.^[4] Among all CPAMs, type 1 is the most common subtype with a frequency higher than 65%; in contrast, type 2 is recognized with non-pulmonary anomalies as a complication. In our patient, in comparing his post-natal CT image to the current illness, the site of pneumonia is consistent with CPAM. As a result, we suspected that pneumonia in our patient may have arisen from the pre-existing structure abnormality.

The timing and necessity of surgical intervention for patients with asymptomatic CPAM are inconclusive currently. If an operative intervention is considered, then pulmonary cyst resection during the infantile period is generally considered to be safe. Previous cases of coexistence of both EDC and type 1 CPAM receiving cystectomy and lobectomy simultaneously all turned out to have a favorable outcome. All previous cases are EDCs complicated by right side type 1 CPAM and subsequently recovered well after surgical removal of both the cyst and the abnormal pulmonary cyst. However, we did not perform lobectomy due to following reasons:

1. the type 2 CPAM without significant space occupation in our patient is not a high-risk lesion and thus is not the direct cause of dyspnea;
2. we show the first case of an EDC complicated by type 2 instead of type 1 CPAM, while there is little possibility for a malignant change in a type 2 CPAM; and
3. early resection of an asymptomatic CPAM still causes concern of potential overtreatment.

Due to the above reasons, we scheduled a regular outpatient follow-up with a 6-month interval for our patient. We also

informed the patients' family that optimal operation should be considered if the patient presented with any complication of CPAM, such as frequent recurrent pneumonia, pneumothorax, or chronic cough.

Children with a critical airway compressed by mediastinal mass are at high risk of anesthetic complications. Orthopnea, upper body edema, stridor, and wheezing are signs of high risk of cardiorespiratory events; although the mortality rate for surgery and anesthesia is quite low. Airway collapse or cardiovascular complications may develop from a previously asymptomatic patient in the process of sedation. Consequently, a pre-operative blind treatment without histological diagnosis is reasonable.^[13] In our case, we pre-medicated the patient with steroids and inhaled bronchodilators for airway maintenance.

5. Conclusions

Children presenting with persistent stridor with trachea deviation who is suspected having a mediastinum mass should undergo further chest CT scan for confirmation. The patient's age, symptomatology, location of mass, and tissue characteristics are helpful information to narrow down the differential diagnosis.^[14] Therefore, EDC should be kept in mind, especially for a child with CPAM.

Author contributions

Conceptualization: Chien-Heng Lin.

Data curation: Ching-Fang Sun, Chieh-Ho Chen, Pin-Zuo Ke, Tzu-Lung Ho, Chien-Heng Lin.

Investigation: Ching-Fang Sun, Chieh-Ho Chen.

Methodology: Chien-Heng Lin.

Resources: Chieh-Ho Chen, Pin-Zuo Ke, Tzu-Lung Ho, Chien-Heng Lin.

Supervision: Chieh-Ho Chen, Chien-Heng Lin.

Writing – original draft: Ching-Fang Sun.

Writing – review & editing: Chien-Heng Lin.

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