Esophageal duplication cyst with hemivertebrae: A case report and literature review

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Abstract

Background: Esophageal duplication cysts (EDCs) are rare congenital anomalies that can be associated with symptomatic spinal abnormalities, but presentations due to EDC symptoms are rarely found in the presence of spinal abnormalities.

Case summary: A 6-month-old infant weighing approximately 5.0 kg presented with a 2-month pulmonary infection and more recent difficulty swallowing and nutritional intolerance that did not improve with medical treatment. Contrast-enhanced chest computed tomography showed a well-defined, mediastinal, homogeneous, low-density cystic mass of 11.9 × 5.5 × 5.1 cm, compressing the liver and bending the trachea forward. Hemivertebrae were present (T4 and T3). Diagnostic laparoscopy was performed, but was converted to open surgery. After ensuring that the cyst was not within the abdominal cavity, thoracotomy was performed, and the cyst was completely resected. Pathophysiologic examination revealed an EDC. The patient recovered well, without symptoms 6 months later.

Conclusions: Overall, noninvasive imaging and diagnostic procedures may not be sufficient to define the exact location of an EDC. Although hemivertebrae were present, they were asymptomatic and did not require treatment; only the EDC induced nonspecific symptoms that disappeared after surgery.

Abbreviations: CT = computed tomography, EDC = esophageal duplication cyst.

Keywords: esophageal duplication cyst, hemivertebrae, infant

1. Introduction

Congenital esophageal duplication cysts (EDCs) are rare congenital anomalies and can be associated with other congenital anomalies such as small intestinal duplication, esophageal atresia distal to the duplication, and tracheoesophageal fistulas. They are often associated with spinal abnormalities, including scoliosis and fusion, which help achieve correct diagnosis. Neuroenteric cysts have been reported to be associated with butterfly vertebra, spina bifida, and hemivertebrae. The incidence of congenital esophageal cysts is estimated at 1:8200, with 2:1 male predominance. Patients with esophageal cysts usually present with pulmonary infection, respiratory distress, difficulty swallowing, and nutritional intolerance due to compression and infection. Most of these cysts are benign, with asymptomatic anomalies that occur during foregut formation; neurological complications are usually the reasons for initial investigation. The preferred treatment is complete surgical resection, and the operation is not difficult. Physicians should be familiar with this disease. We herein report a rare case of a large esophageal duplication cyst with hemivertebrae with an initial presentation of esophageal symptoms.

2. Case report

A 6-month-old Uighur Chinese male infant presented with a history of pulmonary infection for at least 2 months. He had received treatments for pneumonia, using aerosolized second-generation antibiotics. Difficulty swallowing and nutritional intolerance appeared after the onset of pulmonary infection. The infant was born at 38 weeks (3200 g and Apgar score of 9), through cesarean section to a second-time pregnant mother. Antenatal and perinatal histories were uneventful. There was no family history of congenital diseases. On physical examination, the right lung had voiceless percussion, and wheezing could be heard in both lungs. The infant had nutritional intolerance and funnel chest. At admission, chest X-ray was performed, and the patient was diagnosed with bronchopneumonia. The electroencephalogram, limb electromyogram, cardiac ultrasound, blood count, and routine biochemical parameters were all normal. Barium swallow examination did not show substantial luminal narrowing or trace pressure (Fig. 1A). The day after admission, 3-dimensional computed tomography (CT) reconstruction demonstrated a cystic mass located in the posterior mediastinum. This mass displayed well-defined margins and was fluid-filled, but free of air-fluid levels. The cyst was 11.9 × 5.5 × 5.1 cm. The average
wall thickness was 0.5 cm and increased from the extra-pleural space or enterocelia, isolated within the esophagus. The cyst compressed the fore lying trachea, right main bronchus, right inferior lobe (anteriorly), and liver (Fig. 1B). The hemivertebrae were located in the upper thoracic spine (T4 and T3) (Fig. 1C).

Because the cyst was oversized and compressed both the right main bronchus and liver, we performed a diagnostic laparoscopy 7 days upon admission, but this remained inconclusive regarding the exact localization and extent of the cyst. Laparoscopy was converted to open surgery, and the cyst was confirmed to protrude in the abdominal cavity from the thoracic cavity. Then, anterolateral thoracotomy was performed. The cyst was dissected from the adjacent tissues and completely excised. We withdrew about 150 mL of a clear, jelly-like fluid from the cyst. Closed tube drainage was performed for 3 days, and discontinued when chest X-ray showed improvement. The patient was discharged 6 days later when chest X-ray showed improvement of bronchopneumonia, without change in the condition of the spine. The infant recovered uneventfully. Histopathological examination after hematoxylin and eosin staining of the cyst showed gastrointestinal-type mucosa with well-developed muscularis propria and serosa (Fig. 1D).

Since neurological examinations were normal, no treatment for hemivertebrae was undertaken at that time. The infant showed normal growth and development at 6-month follow-up. No X-ray was performed at that time.

Informed consent was obtained from the patient’s parents.

3. Discussion

Esophageal duplication cysts are rare, benign mediastinal masses that occur in the 6th week of embryonic life. They can be associated with symptomatic spinal abnormalities, but presentations due to esophageal duplication cyst symptoms are rarely found in the presence of spinal abnormalities. We reported the
Because the neurological functions were normal and the patient was asymptomatic, no spinal surgery was undertaken. The infant had normal growth and development at 6-month postnatal examination. Complete surgical excision of the cyst was performed successfully during the same operating time.

Only 10% of esophageal cysts interact with the lumen of the esophagus. They can be associated with spinal abnormalities, but the patients usually present with symptoms of spinal abnormalities and are incidentally diagnosed with esophageal duplication cyst after being investigated for respiratory tract symptoms, but this case had no spinal deformity.

Esophageal duplication cysts are rare (incidence of 1:8200). They can be associated with spinal abnormalities, but the patients usually present with symptoms of spinal abnormalities and are incidentally diagnosed with esophageal duplication cyst. Because the neurological functions were normal and the patient was asymptomatic, no spinal surgery was undertaken. The infant had normal growth and development at 6-month postnatal examination.

In conclusion, esophageal duplication cysts are relatively rare and may have no specific symptoms. A combination of multiple imaging and diagnostic procedures may not be sufficient to identify the exact localization of esophageal duplication cysts. Although hemivertebrae were present, they were asymptomatic and did not require treatment; only the esophageal duplication cyst induced nonspecific symptoms that disappeared after treatment. The previous published case was also incidentally diagnosed with esophageal duplication cyst after being investigated for respiratory tract symptoms, but this case had no spinal deformity.

### Table 1

| Case | Sex   | Age  | Country of study | Clinical symptoms                          | Imaging findings                        | Final diagnosis                         | Surgical intervention | Outcome          | Reference               |
|------|-------|------|------------------|-------------------------------------------|----------------------------------------|----------------------------------------|-----------------------|------------------|------------------------|
| 1    | Female | 54 y | Czech Republic   | Dysphagia and a painful swallowing        | $10 \times 4 \times 4 \text{ cm}$ infected cyst with a dense content corresponding to the posterior mediastinum | Inflamed mediastinal duplication cyst    | Partial laparoscopic resection | Favorable       | Zitemory et al. [10] |
| 2    | Female | 21 y | Japan            | Repeated chest pain                       | $2 \times 1.5 \text{ cm}$ cystic lesion   | Mediastinal abscess                     | Thoracoscopy           | Favorable       | Takemura et al. [11] |
| 3    | Female | 12 y | The United States | Shortness of breath                       | $2 \times 2 \text{ cm}$ cystic lesion     | Esophageal duplication cyst             | Robotic-assisted thoracoscopic resection | Favorable       | Obasi et al. [12]    |
| 4    | Male   | 15 y | The United States | History of spina-bifida, hydrocephalus, VP drain, and back pain | $2.6 \times 1.6 \text{ cm}$ heterogeneous echotextured lesion with anechoic component in the lower esophagus | Esophageal duplication cyst             | Surgical excision      | Favorable       | Chaouchy et al. [13] |
| 5    | Male   | 35 y | India            | Dysphagia                                  | $0.5 \times 0.5 \times 0.5 \text{ cm}$ oval-shaped cyst-like tumor located in the extrapleural space | Double esophageal duplication cyst       | En bloc resection       | Favorable       | Zhang et al. [14]    |
| 6    | Male   | 3 y  | China            | Intermittent fever of acute onset and dry cough | $2.1 \times 1.5 \times 2.6 \text{ cm}$ cystic mass in the left posteroinferior mediastinum | Intrathoracic esophageal cystic duplication | Thoracoscopic excision | Favorable       | Mengio et al. [15]   |
| 7    | Male   | 6 mo | Switzerland      | Slowly led with a bottle; otherwise no specific symptoms | Cystic mass measuring $3.5 \text{ mm}$ in proportion to right lung base | Esophageal duplication cyst             | Thoracoscopic excision | Favorable       | Cuch et al. [16]     |
| 8    | Female | Neonate | Poland         | Congenital thoracic cystic mass           | $3.7 \times 2.3 \times 1.5 \text{ cm}$ well-defined cystic lesion | True intrathoracic esophageal duplication cyst (VATS) excision | Favorable       | Ali-Riyami and Al-Sawaf [17] |
| 9    | Male   | 24 y | Oman             | No apparent symptom                       | $9.4 \times 3.1 \times 2.9 \text{ cm}$ in the left paravertebral region | Tubular cystic lesion                   | Thoracic esophageal duplication cyst | Favorable       | Obasi et al. [18]    |
| 10   | Female | 18 y | India            | Fever, cough, and breathlessness           | $3.5 \times 2.3 \times 3 \text{ cm}$ hypodense homogeneous cystic lesion | Esophageal duplication cyst             | Complete surgical excision of the cyst | Favorable       | Sonthalia et al. [19] |
| 11   | Male   | 30 y | India            | Gradually progressive dysphagia           | $2.5 \times 1.3 \text{ cm}$ cystic lesion in distal esophagus | Distal esophageal duplication cyst with gastro-esophageal reflux disease | Left thoracotomy, excision of the duplication cyst and thoracic fundus duplication | Favorable       | Jan et al. [20]      |
| 12   | Male   | 16 mo | United Arab Emirates | Diaphragmatic hernia                      | $11.9 \times 5.5 \times 5.1 \text{ cm}$ cystic mass | Esophageal duplication cyst             | Thorsotomy             | Favorable       | This study          |
| 13   | Male   | 6 mo | China            | Esophageal symptoms                       | $1.5 \times 2 \times 2 \text{ cm}$ cystic mass | Esophageal duplication cyst             | Thoracoscopy           | Favorable       | Tomar et al. [21]    |

### Table 1.

Demographic data, clinical and image findings, surgical interventions, and outcomes in selected cases with esophageal duplication cysts.

- **Case**: Case number.
- **Sex**: Male or Female.
- **Age**: Age in years.
- **Country of study**: Location where the patient was treated.
- **Clinical symptoms**: Symptoms experienced by the patient.
- **Imaging findings**: Description of the cystic lesion as seen on imaging.
- **Final diagnosis**: Diagnosis made after the initial evaluation.
- **Surgical intervention**: Type of surgery performed.
- **Outcome**: Outcome of the surgery.
- **Reference**: Reference for the case study.
medical imaging techniques, such as esophagography, CT scan, and barium swallow examination can assist in diagnosing esophageal duplication cysts and defining their relationships with the neighboring anatomy. Curative surgery is not difficult and is needed to provide strong clinical evidence for diagnosis. Complete surgical excision is typically curative because recurrence is rare.

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