Congenital esophageal diverticulum in a very low birth weight infant: case report and review of literature

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ABSTRACT

Background. A diverticulum is an outpouching of a tubular organ that is classified as congenital and acquired according to the involved layers of the gastrointestinal wall. Congenital true diverticulum has been very rarely seen in neonatal period and it is very difficult to diagnose especially in premature infants.

Case. A male infant was born with birth weight of 1000 g at 28th gestational week, was hospitalized for prematurity and respiratory distress. During follow up intermittent CO₂ retention was observed in blood gases. On the 17th day of hospitalization, esophageal dilatation was detected on X-ray and barium swallowed esophagram showed a saccular pouch on the distal esophagus. The patient was operated on 26th day of life and pathological specimen revealed true diverticulum of esophagus. The patient died due to respiratory failure and septic shock during hospitalization.

Conclusion. To the best of our knowledge this case is the smallest and youngest preterm infant diagnosed with congenital esophageal diverticulum. Prolonged and intermittent CO₂ retention such as in our case can be an atypical symptom of congenital diverticulum and it should be suspected in the differential diagnosis. Congenital esophageal diverticulum may be also seen in extremely preterm infants and can present with unusual symptoms.

Key words: congenital, esophageal diverticulum, newborn, premature.
Case Report

A male infant was born by cesarean section at 28 weeks’ gestation to a 22-year-old mother with Apgar 5 and 7 at 1 and 5 minutes. The birth weight was 1000 g (10-50th centile), birth length was 36 cm (50th centile) and head circumference was 25 cm (10-50th centile). The patient was hospitalized and monitored. Physical examination revealed respiratory distress and prematurity findings. No other congenital anomalies were observed with clinical and radiological findings. Early rescue surfactant was administered. He was given ampicillin and gentamicin therapies for sepsis. He could not be extubated during the first days of life due to the intermittent CO₂ retention that was not compatible with the chest X-ray findings. Although chest radiograms did not reveal reticulogranular image and ventilator parameters were upgraded, there was no dramatic change in CO₂ levels during this period. This condition continued approximately once or twice a day. Ventilatory support status and blood gases changes are summarized in Table I. On the postnatal 17th day, a dilatation at esophagus was seen on chest radiogram (Fig. 1) and barium swallowed esophagram showed a saccular pouch on middle-distal esophagus (Fig. 2). Computed tomography of thorax illustrated a hypodense lesion with cystic characteristics with 30x14x15 mm in diameters forcing lung posteriorly in paraesophageal zone in the left hemithorax. After consultation with pediatric surgeons, esophagogastrroduodenoscopy was performed in the operation room under general anesthesia and no evidence of diverticulum was detected. In addition to the persistence of unexplained intermittent CO₂ retention and persisting dilatation in the mid-esophageal region, the infant was operated on 26th postnatal day of life. After left thoracotomy at 5th intercostal area, grayish-like color mass with 3x1.5x2 cm diameter was visualized between aorta and vertebra in posterior mediastinum left to distal esophagus. The mass was resected and no continuation or opening to any other part of esophagus or mediastinum was observed. The esophagus was primarily repaired and closed. Pathological evaluation of the mass revealed a true diverticulum containing all layers of esophagus. However, after the operation, the patient died on postnatal 34th day due to severe respiratory failure and septic shock. Informed consent was obtained from the parents.

<table>
<thead>
<tr>
<th>Date of Blood Gases</th>
<th>pH</th>
<th>pCO₂ (mmHg)</th>
<th>Ventilatory support status</th>
</tr>
</thead>
<tbody>
<tr>
<td>1st day (Before surfactant therapy)</td>
<td>7.22</td>
<td>64</td>
<td>SIMV mode</td>
</tr>
<tr>
<td>1st day (After surfactant therapy)</td>
<td>7.36</td>
<td>42</td>
<td>SIMV mode</td>
</tr>
<tr>
<td>3rd day</td>
<td>7.25</td>
<td>59</td>
<td>SIMV mode</td>
</tr>
<tr>
<td>7th day (Before extubation)</td>
<td>7.40</td>
<td>38</td>
<td>SIMV mode</td>
</tr>
<tr>
<td>7th day (After extubation)</td>
<td>7.21</td>
<td>92</td>
<td>Nasal CPAP mode</td>
</tr>
<tr>
<td>17th day</td>
<td>7.18</td>
<td>104</td>
<td>HFOV mode</td>
</tr>
<tr>
<td>26th day</td>
<td>7.26</td>
<td>59</td>
<td>SIMV mode</td>
</tr>
<tr>
<td>34th day</td>
<td>6.92</td>
<td>142</td>
<td>HFOV mode</td>
</tr>
</tbody>
</table>

pH: power of hydrogen, pCO₂: partial pressure of carbon dioxide, SIMV: synchronized intermittent mandatory ventilation, CPAP: continuous positive airway pressure, HFOV: high frequency oscillation ventilation.

Discussion

Herein, we described the smallest and youngest preterm infant diagnosed with congenital esophageal diverticulum. The clinical presentation of the case revealed unexplained intermittent respiratory acidosis and respiratory distress that resolved and reoccurred spontaneously. Most of the
Esophageal diverticula are acquired and seen in middle-aged group, it has been very rarely been reported in infants.\(^4\)

Holderman\(^5\) first described a 5-day old infant misdiagnosed with congenital diverticulum in 1927, it was thought to be a class 3 tracheoesophageal fistula. Rush and Stingily\(^6\) reported a newborn with supraclavicular mass causing respiratory obstruction. The autopsy revealed a large congenital cricopharyngeal diverticulum. Also, diverticulum in a newborn was reported by O’Bannon\(^7\) on autopsy including a pouch anteriorly and inferiorly proximal to distal esophagus. In an extended review of literature, Poncher and Milles\(^8\) could not find any examples of congenital esophagus diverticulum in 1933. In a more recent paper, an 8-day old term infant with stridor and poor feeding was diagnosed with upper esophageal diverticulum.\(^9\) However, to our best of knowledge, no preterm infant had a diagnosis of esophageal diverticulum. Therefore, our case represents the smallest preterm infant diagnosed with congenital esophageal diverticulum in the literature.

Development of congenital diverticulum was suggested based on esophageal motor dysfunction by D’Abreu\(^10\) in 1949 and it was manometrically confirmed by Cross et al.\(^11\) in 1961. Kaye\(^12\) reported esophageal motor dysfunction in 12 patients diagnosed with diverticule in 1974. Further theories supporting the development of diverticula were suggested by Ishigami et al.\(^13\) in 1965. Our case had unexplained intermittent respiratory acidosis that did not response to ventilatory therapy and was not compatible with radiological findings. Therefore, motor dysfunction, impaired relaxation and delayed passage in esophagus might have caused increased pressure on the esophagus.\(^12\) The weakness of the muscular layer of esophagus and pouch formation might have resulted in increased pressure against the lungs leading to worsening of pulmonary function. The blood gases might be found normal possibly when the size of the pouch and associated pressure against lungs decreased.

Clinical features are broad for esophageal diverticula. It can be asymptomatic but dysphagia is a common finding. When
symptoms occur, they are likely to be caused by associated underlying motility disorder. Other clinical features associated with esophageal diverticulum include feeding problems, weight loss, regurgitation, stridor, belching, bleeding, and cough. In a recent case report; esophageal diverticulum with bronchoesophageal fistula was diagnosed in case of unexplained cough or recurrent pneumonia. In our case, intermittently increasing CO$_2$ levels were detected as the main and unusual symptom. We suggest that dilatation of esophagus and sac formation may have led to increased pressure on lungs. We also think that it resolved spontaneously when the pressure decreased.

According to adult American Gastroenterological Association guidelines, barium swallowed esophagogram is the gold standard diagnostic method. On barium swallowed esophagogram, diverticulum will be illustrated as a distended, barium filled sac above the diaphragm. We showed the same similar findings in our patient. There was dilated, pouch-like formation seen in the mid-esophageal area. Endoscopy and bronchoscopy may also be useful for both confirming and assessing the degree of esophageal inflammation and obstruction. Carcinomas or other diseases should also be excluded by endoscopy. Although we performed an endoscopy, we could not diagnose the diverticulum. Endoscopy may be dangerous due to increased risk of perforation with misdirection of the scope into the diverticula and is not recommended in case of large diverticula because of incomplete emptying of pouch remnants. In our case, thorax CT showed the diverticulum as a hypodense lesion with cystic characteristics, therefore we suggest to use of other diagnostic tools such as CT in suspected cases. This CT finding can be interrupted with esophageal duplication cyst. But approximately 90 percent of esophageal cysts do not communicate with the esophageal lumen. In our case the lesion originated from the esophageal lumen. We could precisely differentiate this lesion from duplication cyst by pathological evaluation.

The optimal management of these cases are early and prompt surgery. Surgical approach for treatment of diverticulum is mainly myotomy, supported by various studies in which 80-100% of patients had good outcomes. Left-sided thoracotomy is mostly preferred to visualize the esophagus. Our case underwent a left thoracotomy and after the resection of the sac, all esophageal layers were closed anatomically. Although the surgery was successful, the patient died due to problems of prematurity and neonatal sepsis. The prognosis varies according to several factors including the presentation, gestational age, and associated abnormalities. However, the prognosis may be poorer in preterm infants such as in our case. Therefore, we recommend prompt surgery in appropriate conditions after the diagnosis.

The strength of this case can be suggested as the youngest and smallest preterm infant in the literature to be diagnosed and operated on very promptly. Although symptoms were non-specific our findings may represent a clue for both neonatologists and pediatric surgeons. The weak point of this paper is that the problems may have been associated with prematurity and also there was no genetic data about this congenital anomaly.

Esophageal diverticula are frequently acquired and seen in the middle-aged group, it has very rarely been reported in infants and even rarer in premature infants. Clinicians should keep in mind and be aware of this condition in the case of prolonged intubation and extubation failure with prolonged CO$_2$ retention that could not be related to any other situation.

Herein, we reported an unusual case of a true congenital esophageal diverticulum in a very low birth weight premature infant. Although congenital esophageal diverticulum is rare in neonates, the clinical findings may vary from respiratory problems to feeding problems. The main mechanism responsible from variable symptoms may be increased esophageal pressure. In addition to barium graphics, other diagnostic tools such as endoscopy or thorax...
CT should be performed in suspected cases. Prolonged and intermittent CO\textsubscript{2} retention such as in our case can be an atypical symptom of congenital diverticulum. In conclusion, we suggest that congenital esophageal diverticulum should be kept in mind in the differential diagnosis of infants with respiratory and feeding problems even in preterm babies.

REFERENCES