Late presentation of congenital diaphragmatic hernia

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Received: 23 January 2015, Revised: 24 February 2015, Accepted: 3 April 2015

SUMMARY: Ülgen-Tekerek N, Akyıldız B, Özcan A, Ergül AB, Turan C, Özcan S. Late presentation of congenital diaphragmatic hernia. Turk J Pediatr 2015; 57: 642-645.

Late presentation of congenital diaphragmatic hernia is 5-30% of all congenital diaphragma hernia cases. It can present as Morgagni, Bochdalek and paraesophageal hernia. Misdiagnosis can result in significant morbiditiy.

A 17-month-old girl presented with vomiting and abdominal pain. On physical examination, circulatory disturbance, cyanosis, abdominal distantion were present. Her O2 saturation was 60% and she was tachycardic (180 bpm) and tachypneic (58 bpm) with hypotension (60/35 mmhg). Patient's heart and mediastinum were shifted into the right hemithorax on the chest X-ray. Bowel loops in the left hemithorax with air-fluid levels were seen in her plain X-ray and diaphragmatic hernia was seen in her computed tomography examination. She was referred to our center and operated within an hour. Herniated intestinal loops and stomach were observed from about 2 cm diameter defect of diaphragma were repaired primarily. She was extubated in postoperative 4th day.

Late presentation of congenital diaphragmatic hernia may be confused with many situation and is difficult to diagnose without clinical suspicion. Accurate diagnosis and urgent treatment is lifesaving.

Key words: congenital, diaphragmatic hernia, late.

Congenital diaphragmatic hernia (CDH) has been described since the 17th century with a high mortality rate. It depends on incomplete closure of pleuroperitoneal channel during the fetal development[1]. Its estimated incidence has been reported 1/2000–5000 live births[2].

The majority of hernias are seen in the left side (88%), 10% are seen right sided and 2% are seen bilateral[3].

Congenital diaphragmatic hernia usually presents with respiratory distress during the neonatal period, and is associated with pulmonary hypoplasia. The prognosis depends on the degree of pulmonary hypoplasia, pulmonary hypertension and other anomalies[2].

Late presentation of congenital diaphragmatic hernia is 5-30% of all congenital diaphragma hernia cases and it can present as Morgagni (retrosternal defect), Bochdalek (posterolateral defect) and paraesophageal hernia[4]. CDH may cause acute or chronic respiratory and gastrointestinal symptoms. In some cases, gastric volvulus has been reported[5].

Additionally, congenital lung anomalies such as pulmonary sequestration, cystic adenomatoid malformation, congenital lobar emphysema may also accompany[6]. Misdiagnosis can result in significant morbiditiy[7].

In this case report, we wanted to emphasize that congenital diaphragmatic hernia can be seen other than the neonatal period and clinical suspicion is important to identification. Proper diagnosis and treatment is life saving and prevent complications. We report a case of late presenting congenital diaphragmatic hernia treated in our department.

Case Report

The patient is a 17-month-old girl with no remarkable medical history. She was followed-up at another center due to fatigue, fever and antibiotic therapy was started with a diagnosis of tonsillitis. Three days later she presented with vomiting and abdominal pain. Bowel loops in the left hemithorax with air-fluid levels were seen in her direct abdominal radiography (Fig. 1) and diaphragmatic hernia was seen in her computed tomography (CT) examination (Fig. 2). She was referred to our center for urgent operation.

Upon arrival to our department she was intubated. On physical examination, circulatory disturbance, cyanosis, abdominal distantion were present. Her O_2 saturation level was 60% and she was tachycardic (180 bpm) and tachypneic (58 bpm); and her blood pressure was 60/35 mmHg. Patient's heart and mediastinum were shifted into the right hemithorax on the chest X-ray nasogastric tube was administrated and fecaloid drainage was observed. The patient was operated within an hour.

Herniated and strangulated (but not volvulus) intestinal loops and stomach were observed from about 2 cm diameter defect in the central part of the diaphragma. No hernial sac was identified and the defect was repaired primarily. Resection and anastomosis was performed on 45 cm of necrotic small bowel segment. She was extubated on postoperative 4th day and discharged on postoperative 12th day.

Discussion

The majority of CDHs are diagnosed antenatally and presenting with severe respiratory distress during the neonatal period. However, late presentation beyond the neonatal period occurs in between 5 and 30 percent of cases[4]. It can either be acute or insidious with respiratory or gastrointestinal symptoms. Late presenting CDHs may previously have normal chest X-rays. The outcomes of late-presenting diaphragmatic hernias are usually very good, due to good lung development. Our patient also had no health problems earlier and presented with vomiting. CDH diagnosis was made based on abdomen X-ray.

The Congenital Diaphragmatic Hernia Study Group conducted a 10-year study across 30 centers to review the demographics, clinical manifestation and outcomes of late onset CDH.

Of 3098 cases of CDH, only 79 patients presented with late-onset (2.6%). The male to female ratio was 1.8 with mean age at diagnosis of 372 days[8]. Presenting symptoms of late onset CDH could be classified as respiratory (upper respiratory tract infection, pneumonia, respiratory distress, cough, wheezing, etc.), gastrointestinal (vomiting, abdominal pain, failure to thrive, constipation, etc.), both or asymptomatic. Gastrointestinal symptoms predominate in left sided hernias whereas respiratory symptoms are more common in right-sided lesions[8]. This emphasizes, the rarity of the case as our patient was female with age of 17 month at presentation and her left sided CDH presented with gastrointestinal symptoms.

Several theories have been put forward to explain the mechanisms that prevent early presentation. First theory states that the pre-existing diaphragmatic defect is plugged by a solid organ such as the spleen or liver. In the second theory, early herniation is prevented by a 'sac' associated with the abdominal contents. Herniation is subsequently triggered from one or a combination of the following events. A rise in intra-abdominal pressure, a decrease in intra-thoracic pressure, or a rupture of the hernial sac[7]. A new theory has been proposed by Yamamoto et al.[6] to explain the late presentation of early herniation. They reported a case of late presenting congenital diaphragmatic hernia associated with a previous ipsilateral lobectomy for congenital lobar emphysema. They could not explain the case with the traditional theories because of no organs were plugging the hernia and no hernial sac was found. They notified that herniation is prevented by the thick muscular posterior rim of the diaphragmatic defect that overlies the anterior rim. In our case we think that it is more convenient to explain the mechanism of herniation with the last theory.

Yap et al.[9] reported a 17- year-old girl patient of CDH. She presented with left-sided abdominal pain, urinary frequency and vomiting. Urinary tract infection was diagnosed. Since there was no improvement of symptoms, a diagnostic laparoscopy had been made for suspected ruptured ovarian cyst. In the early postoperative

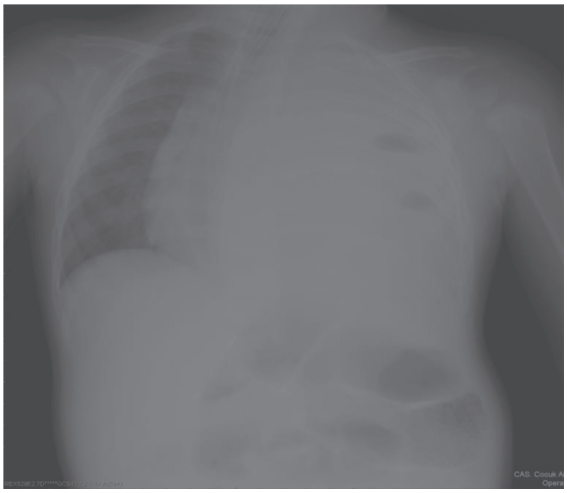

Fig. 1. Bowel loops in the left hemithorax with air-fluid levels were seen in direct abdominal radiogaphy of patient.

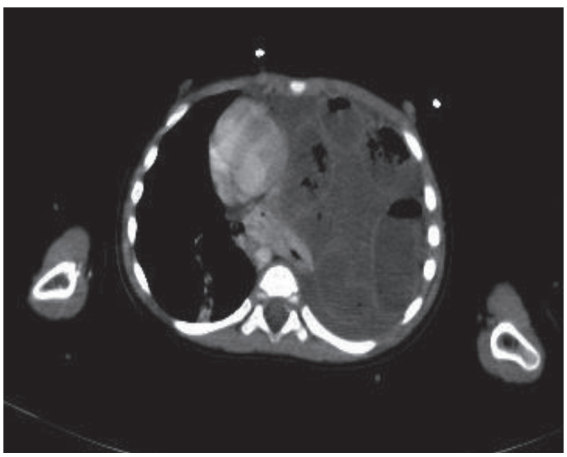

Fig. 2. Diaphragmatic hernia was seen in computed tomography examination.

recovery period, cardiorespiratory distress had occurred and a large intestinal left diaphragmatic hernia was diagnosed. She was operated and discharged a few days after the operation [9].

The prognosis of late-onset CDH is excellent once the correct diagnosis is made. However, making the correct diagnosis is challenging because of its diverse clinical presentations. Clinical suspicion and chest X-ray remain key. Nevertheless, the variety of organs and the size of herniation may make chest X-ray interpretation difficult and late presenting CDH may mimic pneumonia, tumor and diaphragmatic eventration. The hernia may be intermittent and the presence of a normal chest X-ray does not exclude the diagnosis. Further imaging evaluation (e.g. contrast series or CT) is warranted if clinical suspicion is high[7, 10-12].

Following diagnosis, the management of acute presentation of CDH aims to achieve urgent bowel decompression. This is critical to prevent complications such as gastric volvulus, bowel strangulation or respiratory complications[9]. We have applied our patients nasogastric decompression. However resection and anastomosis was performed on 45 cm of necrotic small bowel segment.

Chao et al.[1] Have been reported 15 cases of late presenting CDH from 1987 to 2008. Large bowel herniation was determined in most of the cases (ten cases). Diaphragmatic defect sizes were observed 1.5 to 15 cm and all of the defects were repaired primarly, no mortality was seen. In our patient, ileal loops and stomach have been hearniated from 2 cm diaphragmatic defect and further operation was performed.

In conclusion, late presentation of congenital diaphragmatic hernia may be confused with many situations and is difficult to diagnose without clinical suspicion. Accurate diagnosis and urgent treatment is lifesaving. Early surgical intervention increases the quality of life of the patient by preventing possible complications.

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