A case report and literature review of the late presenting congenital diaphragmatic hernia

Gholamreza Kalvandi^a, Iraj Shahramian^b, Ali Bazi^c, Seyed Mohammad Kazem Razavipour^a and Mojtaba Delaramnasab^c

Late presenting congenital diaphragmatic hernia is a disease associated with defective diaphragm and penetration of different organs into the thoracic cavity. In the present case, a 3-year-old boy was referred to our hospital complaining of acute abdominal pain. No other gastrointestinal symptoms including nausea, vomiting, or constipation were observed. The patient represented no respiratory problems such as dyspnea or respiratory distress syndrome. Radiograph of the thoracic and abdominal cavities showed bowel loops occupying the entire space of the left hemithorax and right-shifted mediastinum. The patient was referred to the pediatric surgery center. The defect was resolved by prompt surgical intervention. A follow-up radiograph within 6 months of

Introduction

Congenital diaphragmatic hernia (CDH) is a multifactorial disorder affecting the thoracic cavity [1–3]. CDH comprises 8% of all congenital disorders, and affects men two times more than women [4]. This condition is distinguished by a defective diaphragm, and results in penetration of the abdominal organs into the chest. CDH is characterized by respiratory failure and high mortality rate in the neonatal period. Late presenting CDH is clinically revealed after 30 days of birth [5]. Late presentation of CDH has been noted in 2.5–20% of CDH patients [5,6]. However, there is an incomplete picture of factors associated with delayed presentation of CDH.

Late presenting CDH shows a different clinical spectrum compared with classic CDH [7]. Regarding this, diagnostic and therapeutic approaches for late presenting CDH are different from neonatal CDH. Late presenting CDH may be suspected in patients with chronic respiratory and digestive symptoms of unknown etiology. In contrast to early diagnosed CDH which often represents as a respiratory distress condition, gastrointestinal problems are more frequently observed in late presenting CDHs [8]. Despite similar pathogenesis, late presenting CDHs [8]. Despite similar pathogenesis, late presenting CDH renders a favorable prognosis if being correctly and timely diagnosed [9]. Correct diagnosis can be made by radiologic evaluation of the chest. Further evaluation for diagnosis can be accomplished by gastrointestinal tract imaging and computerized tomography [10].

Case presentation

A 3-year-old male child with abdominal pain was referred to our hospital. The pain was epigastric with sudden onset at 2 h after a meal. The patient had no signs of abdominal injury, vomiting and diarrhea, fever, abdominal distention, cough, dyspnea, dysuria, frequent urination, or flank pain.

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^aDepartment of Pediatrics, Ilam University of Medical Sciences, Ilam, ^bPediatric Digestive and Hepatic Research Center, Zabol University of Medical Sciences and ^cClinical Research Development Unit, Amir-Al-Momenin Hospital, Zabol University of Medical Sciences, Zabol, Iran

Correspondence to Iraj Shahramian, MD, Pediatric Digestive and Hepatic Research Center, Amir-Al-Momenin Hospital, Zabol, Iran Tel/fax: +98 543 223 2166; e-mail: ir_buper@yahoo.com

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The birth weight of the child was 3400 g. The current weight was 12.5 kg. Respiratory rate was 24 per min, and the pulse rate was 94 per min. Blood pressure and body temperature were 98:65 and 36.8°C, respectively. There was decreased air entry on the left side and cardiac sounds were predominantly heard on the right side. Bowel sound was heard on the left hemithorax. Abdomen was scaphoid and soft without any mass and tenderness. Chest radiograph showed right-shifted mediastinal and bowel loops occupying the entire space of the left hemithorax (Fig. 1). After initial evaluations, the child was referred to a pediatric surgical center. According to the surgery report, abdomen and a part of the stomach had pierced into the diaphragm. The defect was surgically resolved and diaphragm was repaired. The patient was discharged 1 week after the surgery with a stable clinical condition and good appearance. Following 6 months of surgery, a radiograph was obtained which showed no abnormality.

The study was performed considering ethical standards of declaration of Helsinki (https://www.wma.net/wp-content/ uploads/2016/11/DoH-Oct2008.pdf). An informed consent was obtained form the parents before reporting the case.

Discussion

CDH presenting beyond the neonatal period is an unusual phenomenon and a diagnostic dilemma. In CDH patients who are asymptomatic during infancy, either acute abdominal or respiratory symptoms can be the presenting features later in their lives [11]. Late presenting CDH may become evident with either respiratory or gastrointestinal symptoms. The clinical manifestation is variable encompassing respiratory distress or cough-like symptoms, abdominal painful sequela, as well as nausea, diarrhea, and constipation [4,12]. In our patient, the presenting symptom was solely acute abdominal pain, without any other respiratory or digestive

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Chest radiograph of a 3-year-old boy with congenital diaphragmatic hernia.

symptoms. Presenting symptoms in a case of 5 months old male has been described as respiratory along with cardiac distress [7]. In another 17-month-old female, the opening features were vomiting and abdominal pain [13]. Presentations in a 7-month-old female were persistent dyspnea and cough unresponsive to antibiotic administration [9]. In another female patient aged 12 months, CDH was reported with acute respiratory distress accompanied with abdominal pain [14]. In another study on 15 late presenting CDH patients, each of the respiratory and gastrointestinal symptoms were reported in 40%, whereas 20% of the patients showed a combination of these [15]. In addition to such clinical heterogeneity, asymptomatic CDH patients are also seen in 10% of cases [6]. The age of the patients is a feature that can influence clinical presentation in late presenting CDHs. In fact, older patients are more likely to present with abdominal symptoms, whereas younger patients present more commonly with respiratory problems [4,7]. In our 3-year-old patient, abdominal pain was the presenting symptom without any respiratory problems. Nevertheless, a strict relationship between age and clinical presentation has not been established. It is possible that clinical presentation is influenced by other individual, acquired, or genetic determinants yet to be identified.

Regarding the high risk of mortality in late presenting CDH patients, correct and immediate diagnosis of this condition is of critical importance. Timely diagnosis of the defect seems to be the main prognostic factor in CDH [2,15,16]. Delayed diagnosis has been reported in 25% of late presenting CDH with the most common reason has been a nondiagnostic pulmonary radiograph [2,17]. In the study of 15 late presenting CDH during a definite initial diagnosis based on chest radiography was amenable in only 40% of the patients [15]. In another study, chest radiographs were diagnostic in 82.6% of patients with late presenting CDH [18]. In our patient, a combination of chest and abdominal radiography was used for initial evaluation. This showed that the left space of the hemithorax was occupied by the loops of the bowl. The right side of the hemithorax, on the other hand, was replaced by the heart. In the case of an 11-year-old girl who represented with acute abdominal pain, chest and abdominal radiographs showed air-fluid mass in the chest hemithorax alongside with penetration of intestinal loops and mediastinal dislocation [3]. A right-shifted heart and mediastinum within the hemithorax was previously reported in a 17-month-old affected female as well [13]. In another female patient with CDH and age of 12 months, no breath sound was detected in the left side hemithorax which was stuffed with right-shifted mediastinum [14]. Overall, confusing chest radiographs resembling other acute pulmonary disorders such as pneumonia, pleural effusion, or pneumothorax may occur in patients with late presenting CDH [19]. Although observing the gastrointestinal volvulus structure in imaging studies of chest is a helpful feature - as for the case reported in present study - a definite diagnosis can be made by imaging of the abdomen indicating dislocation or absence of gastric bubble [20]. In suspected patients, performing a computed tomography of the chest can further provide valuable information in late presenting CDH [15].

Our patient was a case of left-sided late presenting CDH. The location of the defect is important as the diagnosis of right-sided defects may be missed because of the blockage of the defect by the liver [6]. Left-sided, right-sided, and bilateral disease have reported with frequencies of 64, 26, and 10%, respectively, in a previous study [2]. In other reports, 90% of CDH cases showed left-sided disease [5,9]. In a review study on 349 patients with late presenting CDH, chest radiography was diagnostic in half of the patients with left-sided disease and 44% of patients with right-sided herniation [19]. Pathological evidences suggestive of a liver hypotrophy may be representative of rightsided CDH [21]. It is noted that right-sided CDH cases are more prone to respiratory symptoms, as liver blocks the entrance of the abdominal organs into the thoracic cavity and therefore no gastrointestinal symptoms are formed [9]. In line, our participant was a left-sided case representing with a gastrointestinal problem (i.e. abdominal pain).

Conclusion

It is of critical importance to differentiate late presenting CDH from other potential causes of acute abdominal pain. Timely diagnosis and immediate surgical intervention are inevitable in order to reduce the mortality rate. This can be amenable by a combination of chest and abdominal imaging studies which render a reliable method for the diagnosis of late CDH.

Conflicts of interest

There are no conflicts of interest.

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