Congenital diaphragmatic hernia with late discovery, a case report

ABSTRACT

Congenital diaphragmatic hernia is a condition where the abdominal contents protrude into the thoracic cavity due to a defect in the diaphragm. It is most commonly diagnosed in the prenatal or neonatal period and often leads to severe acute respiratory distress in newborns.

However, the late-onset form of this condition beyond the neonatal period is rare and can often be misdiagnosed, leading to delayed treatment. This can be due to either the late onset of initial symptoms or an asymptomatic form that is incidentally discovered during a chest X-ray beyond the neonatal period.

In this report, we present the case of an infant born from a well-monitored pregnancy, delivered by Caesarean section, with a normal birth weight of 3200 grams and a normal Apgar score. The infant was discharged after 24 hours and remained healthy at home.

At 6 months of age, the infant presented with acute viral bronchiolitis, and a chest X-ray revealed a congenital diaphragmatic hernia with late-onset symptoms. The patient was stabilized and referred to surgery for treatment after the successful management of bronchiolitis.

Key words: Diaphragmatic hernia, congenital diaphragmatic hernia with late discovery, chest radiography.

1. INTRODUCTION:

Congenital diaphragmatic hernia (CDH) occurs when the pleuroperitoneal duct fails to close properly during fetal development. It affects approximately 1 in 3000 live births, with most cases being diagnosed prenatally or neonatally due to severe, life-threatening respiratory distress.

The neonatal form of CDH has a high mortality and morbidity rate. However, the late-onset form of CDH, which accounts for 5 to 25% of cases, has a better prognosis and less severe side effects [1]. This is because pulmonary hypoplasia is often less severe or absent in these cases [2]. When diagnosed earlier, the prognosis for late-onset CDH is usually favorable.

2. PRESENTATION OF CASE:

The reported case involves a 6-month-old male infant from a non-consanguineous marriage. The pregnancy was well attended and carried to term, with the infant delivered vaginally and weighing 3200 grams, with a normal Apgar score. After being discharged from the hospital 24 hours later, the infant remained healthy at home and had no history of trauma or falls.

However, the infant later presented with febrile respiratory distress, preceded three days earlier by viral prodromes. On clinical examination, the infant was found to be conscious, tonic, and normo-colored, but with a staturopendental delay of -2 standard deviations (weighing 4 kg and measuring 56 cm in height). The infant was also polypneic, with a respiratory rate of 62 cycles per minute, tachycardic at 126 beats per minute, and with an SO2 level of 92% in room air. Signs of respiratory struggle, such as sub-costal pulling, supra-sternal pulling with nasal wing flapping, and bilateral snoring and sibilant rales on auscultation, were also observed.

A chest X-ray showed right apical atelectasis with left-sided colothorax [Fig. 1], and a thoracic radiography of profile revealed the colothorax in anterocardiac [Fig. 2].

Paraclinical workup found a normal blood count and negative C-reactive protein, but respiratory multiplex polymerase chain reaction (PCR multiplex) revealed a respiratory syncytial virus (RSV).

The infant was diagnosed with acute viral bronchiolitis on a congenital diaphragmatic hernia terrain, and was treated with nasal decongestion, tracheobronchial aspiration, oxygen therapy, and good hydration.

The infant's condition improved favorably, and the patient was stabilized before being referred to the pediatric surgery service for surgical management.

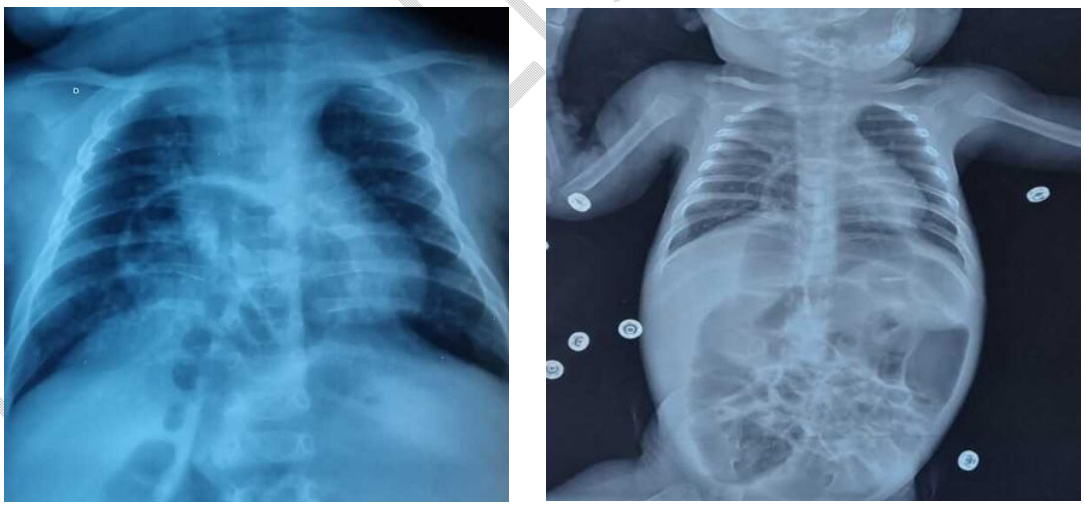

Fig.1 (a.b). A frontal chest X-ray was performed and found a right apical atelectasis with a left-sided colothorax.

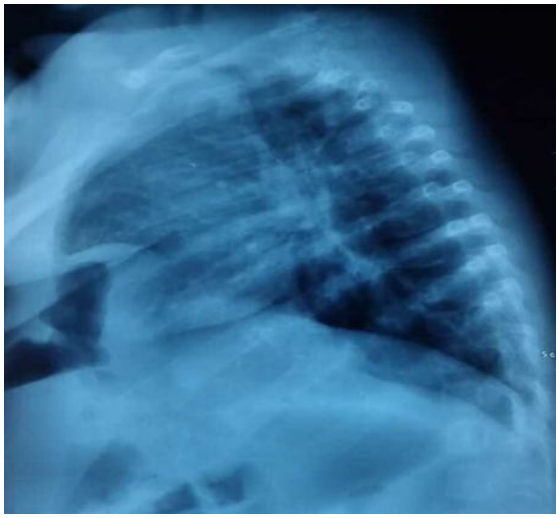

Fig.2. The lateral chest X-ray showing colothorax in an anterior location to the heart.

3. DISCUSSION:

Congenital diaphragmatic hernia (CDH) is an inherited disorder characterized by abnormal growth of the diaphragm, which affects one in 3000 newborns and has an overall survival rate of 67%. This condition is caused by a diaphragmatic tunnel that allows peritoneal viscera to protrude into the pleural cavity [3]. It is most commonly associated with neonatal respiratory distress, however, late-presenting CDH has lesser side effects and a better prognosis [1] due to milder or even absent pulmonary hypoplasia [2]. It represents 5 to 25% of cases, with a male-female ratio of about 2:1.4.

Its diagnosis is difficult due to its insidious onset [4], and factors associated with this late presentation are poorly characterized [5]. Prolonged respiratory and gastrointestinal symptoms may be secondary to this etiology.

Due to its milder and more complex clinical presentation, this type of CDH poses a significant diagnostic challenge [6]. It is often established using chest radiography, and sometimes radiological diagnosis is difficult, which can lead to misdiagnosis. The hernia may simulate lower lobe pneumonia, diaphragmatic eventration, pneumothorax, pleural effusion or even a diaphragmatic mass [7, 8], and may even appear normal due to temporary obstruction of the diaphragmatic discontinuity by the liver or spleen. Associated with a nonspecific clinical presentation, this can delay diagnosis. In these cases, contrast studies of the gastrointestinal tract or thoraco-abdominal CT scan may aid in diagnosis [9].

The content of the hernia is associated with the location of the defect. In the literature, the colon, small intestine, stomach and spleen have successively herniated in cases of left-sided CDH. The liver, small intestine and colon have been herniated in that order in cases of right-sided CDH [5]. In our case, due to the absence of intestinal malrotation, only organs adjacent to the defect or relatively mobile organs, such as the small intestine and transverse colon, were herniated.

The main complications of diaphragmatic hernias, aside from intrathoracic compressive consequences, are intestinal obstruction, hernia strangulation and perforation, which may present as peritonitis or mediastinitis.

Acute gastric volvulus associated with CDH is extremely rare but can have life-threatening complications. It is associated with elongation or absence of two of the four ligaments of the stomach with connection to the left diaphragm. There are three types of gastric volvulus: organo-axial, mesentero-axial or combined. It is often misdiagnosed as pneumothorax, pneumonia with cavitation, pleural effusion or pneumatoceles [10, 11, 12], and initial chest radiographs show a large air bubble with fluid level in the left side of the chest. This common error often leads to iatrogenic gastric perforation due to thoracic drain placement. And the increased intra-abdominal pressure can result in fatal outcomes, such as gastric strangulation, ischemia, perforation, pancreatitis, peritonitis, shock and death, with a mortality rate of 80% [11, 13].

Early surgical intervention is necessary to prevent these complications. Rapid diagnosis is crucial to avoid disruption of the child's development and potentially life-threatening conditions. When surgical repair is performed promptly, the prognosis is generally favorable [14].

4. CONCLUSION:

Congenital diaphragmatic hernia in neonates is a well-recognized condition, but its presentations beyond the neonatal period can vary, leading to clinical and radiological misdiagnoses. As such, it is important for all pediatricians and pediatric surgeons to be aware of the possibility of late-onset congenital diaphragmatic hernia in their routine practice.

Early diagnosis of this condition is crucial and can be achieved through thorough physical examination and proper interpretation of imaging. This early diagnosis allows for early management, which in turn can reduce the possible risks and complications associated with the condition. Early surgical correction is the most effective way to treat this condition and can save lives.

Therefore, it is important to increase awareness of this condition and its potential presentations beyond the neonatal period to ensure timely diagnosis and treatment. By doing so, we can improve patient outcomes and prevent unnecessary morbidity and mortality.

5. REFERENCES:

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