Acute respiratory distress for late-presenting congenital diaphragmatic hernia

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Congenital diaphragmatic hernia (CDH) presents a wide spectrum of anatomical variants and clinical pictures depending on the topography and dimensions of the diaphragmatic defect and on the patient age. Most CDH cases acutely present with tachypnea, cyanosis, and respiratory failure within the first minutes to hours of life. Despite significant advances in neonatal medicine, this congenital anomaly still presents a high mortality rate, especially for associated malformations.

On the other hand, there is a rare subset of CDH patients who present outside the neonatal period. The most common symptoms of late-presenting CDH include recurrent pulmonary infections, dyspnea, wheezing, abdominal pain, failure to thrive, vomiting, diarrhea and anorexia. Although late-presenting CDH generally presents good prognosis after early surgical correction, misdiagnosis is quite frequent because of its wide spectrum of clinical manifestations.

The following case report describes a six-month-old infant presenting with acute respiratory distress and vomiting caused by late-presenting left-sided CDH.

Key words: respiratory distress, congenital hernia, late-presenting, diaphragm.

In surgical causes of respiratory distress in neonates, the underlying mechanisms include airway obstruction, pulmonary collapse or displacement and parenchymal disease or insufficiency; one of the most common causes is congenital diaphragmatic hernia (CHD), with an incidence of 1:2500-5000 live births[1]. CDH represents a protrusion of abdominal viscera through the diaphragm with or without hernial sac. In most CDH cases, visceral hernia occurs posterolaterally through Bochdalek's foramen, which is 20 times more frequent than the anterior CDH through Morgagni-Larrey's foramen (only 1-5% of CDH patients)[1-2]. The presence of abdominal viscera in the chest during fetal life prevents normal organ development, mainly inducing lung hypoplasia. The onset of symptoms depends on the viscera volume in the chest and the degree of pulmonary hypoplasia. A history of polyhydramnios is noted in approximately 80%, and prematurity is frequent[1-2].

Usually, immediate postnatal presentation of CDH is seen, while less severely affected newborns may reveal it beyond the first few hours of life in 10-20% of CDH cases. Cyanosis, tachypnea, grunting and retraction are commonly seen. On physical examination, the abdomen is scaphoid, the chest anteroposterior diameter is increased, a mediastinal shift may be noted, and breath sounds are absent on the affected side while bowel sounds may be heard in the chest[1].

On the opposite side of the CDH clinical spectrum, there is an uncommon subset of patients who present symptoms beyond the neonatal period: these cases with late-presenting CDH represent 10-13% of all CDH cases[3-7]. The symptoms in this late-onset group are non-specific and may commonly include recurrent chest infections, failure to thrive, vomiting, diarrhea, anorexia and abdominal pain. For these reasons, late-presenting CDH is quite frequently misdiagnosed.

Here, we describe an unusual case of a six-month-old infant admitted for vomiting crises accompanied by hyporeactivity, paleness, polypnea and tachycardia due to late-presenting left-sided CDH.

Case Report

A six-month-old boy was admitted to the Pediatric Department with acute progressive respiratory distress and intense vomiting.

Gestational history was unremarkable, particularly in relation to maternal hemorrhage, diabetes, hypertension, trauma, drugs, presence of oligohydramnios or polyhydramnios, and Rh or blood group incompatibility. He was born adequate for gestational age at 32 weeks after spontaneous vaginal delivery (weight 2,400 g, length 46 cm, head circumference 33 cm, Apgar scores 7 and 6 at 1 and 5 minutes after birth). At birth, he was suddenly admitted to the Neonatal Intensive Care Unit for light respiratory distress (neonatal wet lung syndrome)[8], which required continuous positive airway pressure therapy for less than 20 hours. Physical examination did not reveal any pathologic or dysmorphic sign. A chest X-ray was performed without evident signs of anomalies (Fig. 1A). At 7 days of age, the boy was discharged in good health.

The postnatal and infant period was uneventful except for one vomiting episode at two months of age. His growth and development milestones were normal. There was no history of cyanosis, vomiting, diarrhea, constipation, irritability or difficulty in breathing after meals.

At six months of age, the child was admitted to the Pediatric Emergency Unit with acute progressive respiratory distress and intense vomiting. His weight was 6.2 kg (below the 10th percentile), length was 64.0 cm (25th percentile), and head circumference was 42.5 cm (50th percentile). On examination, he appeared ill and wasted. The paleness was accompanied by hyporeactivity, polypnea (respiratory rate 55 beats per minute with 89% blood oxygen saturation) and tachycardia (pulse rate of 160/min, blood pressure of 80/60 mmHg). His body temperature was 37.6°C. There was chest overdistension with greatly increased anteroposterior diameter and with intercostal and subcostal indrawing. The trachea was displaced to the right, and the apex beat and heart sounds were best detected on the right side of the chest. Chest movements and breath sounds were markedly reduced over the left chest but percussion note was bilaterally resonant. Other systems were normal. The initial arterial blood gas while breathing oxygen via a face mask showed particularly mild hypoxia (pH 7.33; pO_2 75 mmHg; pCO_2 36 mmHg; HCO_3 22 mEq/l; Hb 10.0 g/dl).

The chest X-ray demonstrated a well-defined circled bulky aerial content in the left hemithorax, which was presumed to be an intestinal ansa, driving the mediastinum and tracheal axis to the right side of the chest (Fig. 1B). Further radiographic evaluation, which included barium contrast study and sonography, confirmed the suspicion of left-sided posterolateral diaphragmatic hernia with intra-abdominal stomach and wandering spleen. No other malformations were detected in clinical and laboratory investigations.

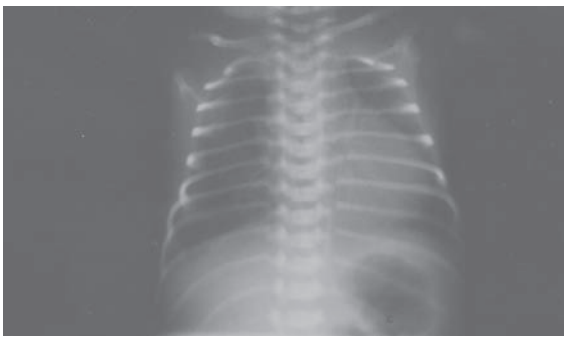
After the initial emergency care, it became necessary to admit the infant to the Pediatric Surgery Unit because surgical intervention was deemed appropriate. By left subcostal abdominal incision, the viscera were lowered into the abdominal cavity, the diaphragmatic defect was fixed by nonabsorbable sutures, and a thoracic drain tube was placed.

A postoperative chest X-ray showed mediastinum realignment with total expansion of the right lung and partial expansion of the left (Fig. 1C). The patient made an uneventful postoperative recovery and remained well at follow-up examination, demonstrating the progressive expansion of the left lung.

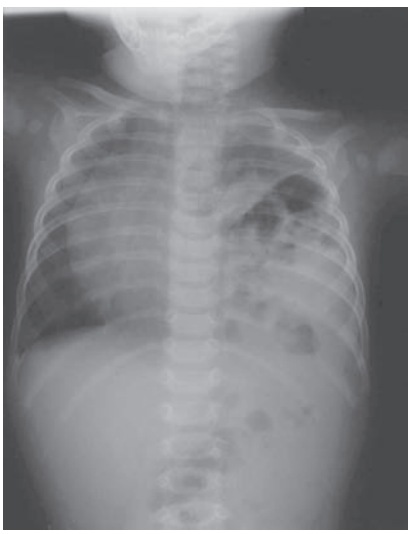
After the local Institutional Review Board approval, informed consent was obtained from both parents prior to preparation of this case report manuscript.

Discussion

Congenital diaphragmatic hernia is a well-known cause of severe respiratory distress in newborns, and is associated with a high mortality rate. There is a milder form of CDH that does not present symptoms until later in life. Fewer than 450 patients with late-presenting CDH were reported in the literature as of December 2006. Today, late-presenting CDH is regarded by pediatric surgeons merely as a clinical variant of CDH. Apart from the principal anatomical



(a)



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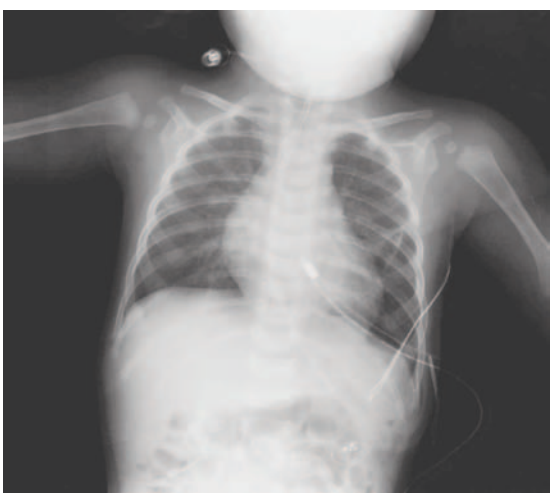

(c)

Fig. 1. (A) Chest radiograph at birth did not show signs of congenital diaphragmatic hernia; (B) Pre-operative chest radiograph revealed gut ansae in the left hemithorax; (C) Post-operative chest radiograph shows the realignment of the mediastinum and the cardiac shadow.

feature of diaphragmatic defect, neonatal and late-presenting CDH significantly differ in some intriguing aspects (Table I).

First, the incidence of neonatal CDH ranges from 0.08 to 0.45 per 1,000 births[1]. The explanation for this variation in incidence is most likely under-diagnosis related to early deaths among neonates who are severely affected. Taking into account the larger series published in the last two decades, the occurrence of late-presenting CDH can be estimated to be 10-13% of all CDH cases[3-7]. Among late-presenting CDH cases, unilateral posterolateral (i.e. though Bochdalek's foramen) defects constituted more than 96%, with 79.4% and 20.6% left- and right-sided hernias, respectively[9]. Such a distribution of anatomical variants corresponds closely to neonatal CDH[10-11].

Second, most neonatal CDH studies have found an equal representation of genders, although a 1.25 male-to-female ratio was reported in one large population-based study[12]. On the contrary, in 349 late-presenting CDH children recently reviewed, the male-to-female ratio was close to 2:1[9].

Third, there is a reported incidence of 40-50% of other malformations in association with neonatal CDH[13], the most common of which are those involving the central nervous system. Therefore, CDH can be seen as an isolated defect with multiple other anomalies, as recognizable nonchromosomal syndrome, or as chromosomal defect (Turner's syndrome, Fryns syndrome, trisomy 13 and 18)[13]. Congenital anomalies, especially those of the heart, are the most common cause of neonatal death (1.7 per 1,000 births), while neonatal CDH may account for 4% to 10% of these deaths[1].

An important epidemiological feature of late-presenting CDH is the very low incidence of major associated anomalies, which stands in great contrast to neonatal hernia[10,13,14]. This fact seems to indicate that neonatal and late-presenting CDH do not share the same pathogenetic factors involved in the formation of the diaphragmatic defect. In addition, lung hypoplasia noted uniformly in newborns affected by CDH -although reported to be mild or moderate in some patients with the late-presenting variant- is without a doubt not a constant feature in this latter group[15].

Table I. Clinical Differences Among Neonatal and Late-Presenting CDH

	Neonatal CDH	Late-presenting CDH
Incidence	0.08-0.45 per 1000 births	10-16% of all CDH
Male-to-female ratio	1:1	2:1
Other malformations	in 40-50% of CDH cases	very low
Symptom onset	acute	acute or chronic
Symptom type	mainly respiratory	respiartory and/or gastrointestinal
Pathologic chest X-ray	in 95% of CDH cases	in 84% of CDH cases high
Survival rate	43.5% of CDH cases	high

CDH: congenital diaphragmatic hernia.

The anomaly underlying CDH is the failure of normal diaphragm development during embryogenesis[2]. Between the fourth and sixth weeks of gestation, failure of formation or fusion of pleuroperitoneal membranes results in a posterolateral diaphragmatic defect that is often referred to as Bochdalek's foramen[16]. This defect occurs five times more frequently on the left side than the right, probably because of earlier closure of the right pleuroperitoneal canal than the left. If the pleuroperitoneal canal remains open when intestines return to the abdomen from the umbilical cord during the tenth week, abdominal viscera move freely into the thoracic cavity. If the pleuroperitoneal canal closes but fails to become muscularized, a hernia with sac results, as seen in 10-15% of patients who have CDH[16]. Chest radiographs of neonatal CDH show air-filled gastrointestinal loops in the chest cavity with non-visualization of the diaphragmatic margin, mediastinal shift and relative paucity of abdominal gas[1]. A small portion of the ipsilateral lung may be visible above the expected position.

On the other hand, Wiseman and MacPherson[17] suggested an "acquired" nature of hernia in children who showed late symptoms and had previously normal diagnostic studies. Therefore, late-presenting hernia may follow a two-hit pattern. The first event must be related to disturbances in the development of the embryonic diaphragm with the formation of a posterolateral defect as potential hernial ring. The second event consists of displacement of intra-abdominal viscera into the thoracic cavity. However, even occurrence of the second hit is not a prerequisite for immediate clinical manifestation of this anomaly. This hypothesis has been supported by other authors[3,7,18] and by chest X-rays potentially relevant to the diagnosis, which were reported as normal in 16% of cases[15]. A normal chest radiograph delineates intrathoracic anatomy before the acquired herniation takes place in postnatal life. For example, our case with normal chest radiograph at birth and the historic absence of specific recurrent symptoms since infancy supports this hypothesis. However, it is unclear whether such herniation may occur intermittently or whether the viscera remain fixed in their intrathoracic position, once displaced[15].

Fourth, late-presenting CDH is characterized by a wide clinical spectrum. CDH patients diagnosed after infancy may present acute, chronic symptoms or may even be completely asymptomatic. More than 80% of infants with late-presenting left-sided CDH presented acutely[9]. The frequency of acute manifestation of left CDH diminishes significantly in older patients, being almost equal to the number of chronic presenters[9,14]. On the contrary, partial liver displacement, which occurs in most right-sided CDH children, produces a clinical picture of chronic complaints[9,19]. It is possible that partial liver herniation may block the further herniation of hollow viscera, preventing the development of gastrointestinal symptoms[14].

This clearly contrasts neonatal posterolateral hernia manifesting itself uniformly with acute respiratory distress[1]. Furthermore, younger patients with late-presenting CDH more frequently show respiratory rather than gastrointestinal symptoms. This is clearly seen in the right CDH group, in which most children have respiratory symptoms only[9,14]. However, older patients, regardless of the defective side, presented higher frequency of gastrointestinal rather than respiratory symptoms[19]. Therefore, none of the symptoms of CDH in an older child should be regarded as pathognomic for late-presenting CDH. Even in an affected infant, the absence of respiratory symptoms should not exclude late-presenting CDH from the differential diagnosis as a potential underlying pathology.

In late-presenting CDH, the most important factor responsible for acute or chronic presentation seems to be the type of herniated viscera[2]. Acute presentation may be due to rapid visceral displacement into the chest or rapid distension of previously herniated hollow viscera in most cases. Chronic symptoms result from longstanding compression of the ipsilateral lung. In such pathological situations, lung compression leads to respiratory distress, or incarceration of the stomach or bowel within a relatively narrow hernial ring producing predominantly gastrointestinal symptoms. Rapid displacement of intra-abdominal organs may lead to sudden cardiorespiratory arrest, which supports the hypothesis of the acquired nature of herniation[17].

Late-presenting CDH is generally considered as a benign condition due to its good prognosis[7,10,20-21]. Such a view is undoubtedly justified when the prognoses for neonatal CDH and late-presenting CDH are compared[19,22]. In population-based studies, the survival rate of neonates who have CDH is 43.5%[1]. Recently, Baglaj[9] suggested that intra-thoracic displacement of the stomach is associated with the highest risk of a complicated, or even fatal, course of late-presenting CDH. Therefore, it may be clearly concluded that late-presenting hernia is an unequivocal indication for operative treatment as soon as its diagnosis has been established.

In conclusion, late-presenting CDH is a very intriguing defect with a wide spectrum of clinical manifestation. All pediatricians and pediatric surgeons should be aware of this condition in their routine practice.

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