ORIGINAL ARTICLE

LATE PRESENTING CONGENITAL DIAPHRAGMATIC HERNIA – A CASE SERIES

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Abstract

With an incidence of 1 to 2500 to 1 to 5000 births, congenital diaphragmatic hernia (CDH) can be a life-threatening condition. In 5-25% of the cases, CDH can prsent at older ages with non-specific signs and symptoms. The aim of our study was to assess the clinical aspects, the diagnosis and management of the patients with this affliction as well as increase the level of awareness on late presenting CDH. Five patients older than 1 year of age at the moment of diagnosis of CDH were included in the study. The patients presented either for respiratory symptoms, or gastrointestinal one. In all the cases, during initial assess, plain chest or abdominal x-rays were performed leading to the diagnosis of congenital diaphragmatic hernia. After further imaging studies (CT scans, ultrasound) all the patients underwent surgery for defect closure, 3 of them by classic approach, 1 by laparoscopy and 1 by thoracoscopy. Three patiens had left diaphragmatic defect and 2 had anterior diaphragmatic defect. We extensively present the case of a 14 year-old female patient with posterolateral diaphragmatic defect, in which gastric necrosis was found during surgery, leading to partial gastric resection and esogastric anastomosis. In this case the postoperative evolution was critical and the patient died in the 9th day postoperatively. The evolution of the other 4 patients was uneventful.

Keywords: *congenital diaphragmatic hernia; late presentation*

Introduction

The incidence of congenital diaphragmatic hernia (CDH) is 1 to 2500 to 1 to 5000 births [1]. Usually it is antenatally diagnosed by ultrasound and is a life-threatening condition, due to its' impact on the pulmonary tract development. However, in 5-25% of the cases [2] CDH is diagnosed at older ages, either incidentally or due to non-specific symptoms which can have either an acute or an insidious onset.

The diaphragmatic defect can be anterior or posterolateral (on the left side, on the right side or bilaterally) and most of the children associate other anomalies.

The aim of our study was to assess the clinical aspects, the diagnosis and management of the patients with late presenting CDH and also to increase the level of awareness about this anomaly of which diagnosis should be taken into consideration in any child presenting with respiratory or gastrointestinal symptoms. In our study, we included 5 patients older than 1 year of age at the moment of diagnosis of CDH. The following data were analysed: age, gender, signs and syptoms at presentation, imaging

investigations, intraoperative discoveries and outcome.

Presentation of the cases

We included in our study 5 cases of late presenting CDH. The study included 3 male and 2 female patients. The mean age at presentation was 6,8 years-old. Only 3 of the patients presented with medical history, as follows: one male presented recurrent airway tract infections, one female presented recurrent abdominal pain and one female underwent appendectomy 1 year before the presentation. At the moment of presentation 3 of the patients had respiratory symptoms (such as shortnes of breath or cough) while 2 of them had abdominal pain, from which one had also multiple vomiting episodes (Table 1).

No.	Age (years)	Gender	History	Clinical aspect at presentation
1	1	M	Recurrent airway tract infections	Respiratory symptoms
2	2	M	None	Respiratory symptoms
3	5	M	None	Respiratory symptoms and fever
4	10	F	Recurrent abdominal pain	Abdominal pain
5	14	F	Appendectomy 1 year before	Abdominal pain and vomiting

Table 1 - Age, gender, medical history and clinical aspect at presentation of the late presenting CDH patients in our study

No.	Age (years)	Gender	Initial imaging	Additional imaging studies
1	1	M	Plain chest x-ray	Thoraco-abdominal US
2	2	M	Plain chest x-ray	Thoraco-abdominal CT scan
3	5	M	Plain chest x-ray	Thoraco-abdominal CT scan
4	10	F	Abdominal and plain chest x-ray	Thoraco-abdominal CT scan
5	14	F	Abdominal x-ray and abdominal ultrasound	Plain chest x-ray, thoracic US and thoraco-abdominal CT scan

Table 2 - Initial imaging and additional imaging studies performed for the 5 patients

No.	Age (years)	Gender	Surgery	Anatomical defect	Outcome
1	1	Male	Classic	Anterior	Uneventful
2	2	Male	Laparoscopic	Anterior	Uneventful
3	5	Male	Thoracoscopic	Posterolateral	Uneventful
4	12	Female	Classic	Anterior	Uneventful

Table 3 - Surgical approach, anatomical defect and the outcome of the 5 patients

The first 3 patients (Table 1), after the primary clinical examination performed by the pediatrician, they underwent a plain chest x-ray and the diaphragmatic hernia was revealed. In the case of the patient no. 4, the pediatric surgeon examined her in the emergency room and an abdominal and thoracic x-ray were performed, which showed the diaphragmatic hernia. All of the patients were admitted to the pediatric surgery department and further imaging studies were performed for the confirmation of the diagnosis (see Table 2).

Due to its' particularities, the 5th case in our series will be presented in details as it follows. A 14 year-old female presented to the emergency room with diffuse abdominal pain and multiple vomiting episodes in the course of 3 days prior to the examination. She had a history of appendectomy. At that time, a chest x-ray was performed, showing no abnormalities. After the initial clinical examination in the emergency department, an abdominal x-ray and ultrasound were performed, neither of them being relevant. Due to her general state she was admitted to the pediatric surgery department, where under

thorough supervision, she developed respiratory symptoms. The patient was referred to the radiology department for a chest x-ray, where she suffered a cardiac arrest. She was resuscitated and admitted to the intensive care unit. After she was stabilized, additional imaging studies were performed (Table 2.) showing the diaphragmatic hernia. Emergency surgery was performed after that.

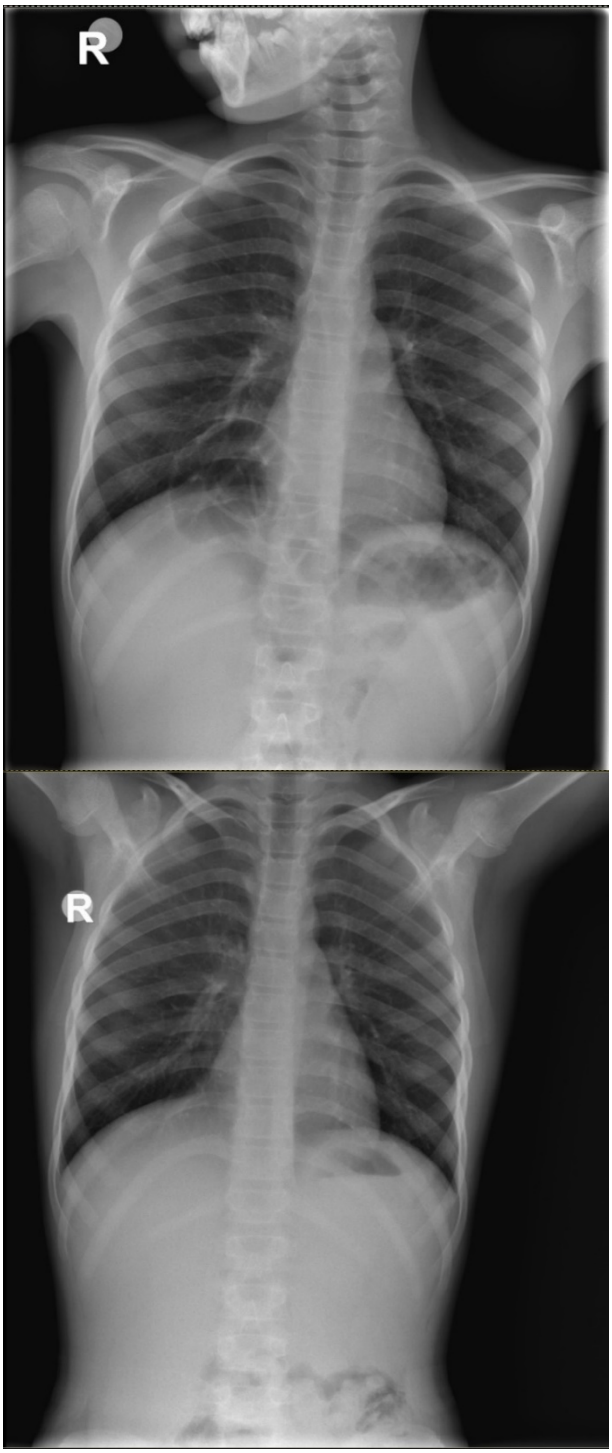

Figure 1 - Pacient no. 5 – chest x-ray before (up) and after (down) surgery

During surgery, the team found a left diaphragmatic posterolateral defect, complete ascension of the stomach in the chest cavity with important gastric necrosis and ascension of spleen which was normal. Using a classic approach, the team performed a subtotal gastrectomy with esogastric anastomosis and closure of the diaphragmatic defect. Postoperative evolution was critical and despite all the efforts the patient died in the 9th day postoperatively.

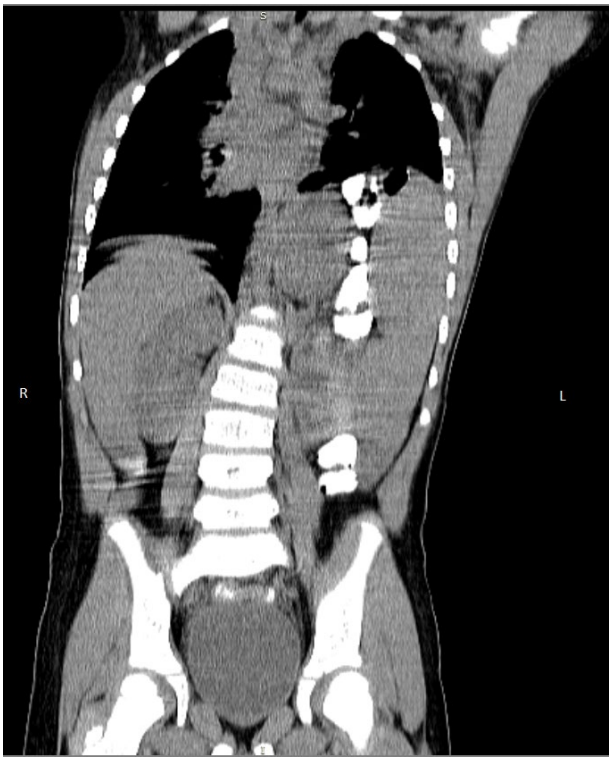

Figure 2 - Thoracic, abdominal and pelvic CT scan of patient no. 3

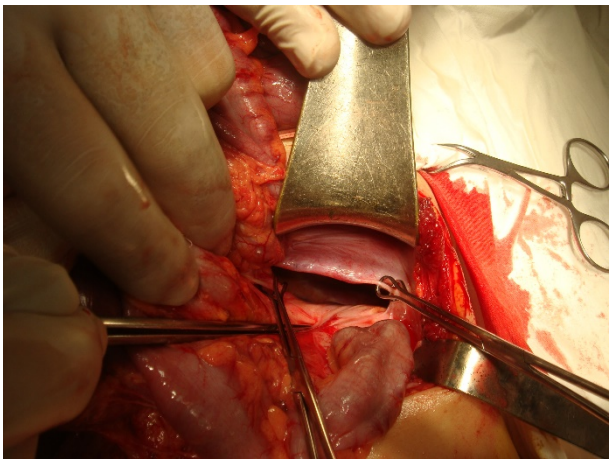

Figure 3 - Example of left posterolateral diphragmatic defect in an incidental diagnosed CDH

All the other 4 patients (Table 3) underwent surgery, 2 of them by classic approach, 1 by thoracoscopic approach and 1 by laparoscopic approach. According to intraoperative findings 3 of our patients had an anterior diaphragmatic hernia, while one of them had left sided posterolateral hernia. All four patients had an eventful postoperatively evolution being discharged in good general state. Median hospital stay was 9 days.

Discussions

Late presenting CDH is an uncommon condition. Coran et al. consider that 10-20% of the patients with CDH will present at older ages [1] while Sanh et al. consider that 5-25% of the children with CDH will present at older ages [2].

The diagnosis of late-presenting CDH is difficult due to many nonspecific signs and symptoms. Three of the patients in our study presented with respiratory symptoms and the others with abdominal pain. According to literature, nonspecific signs and symptoms as dyspnea, tachypnea, cough, wheezing, abdominal pain, vomiting, and are seen in patients with late presenting CDH [3]–[7]. Mei-Zahav et al. split 22 patients with late-presenting CDH in his study into 3 groups. In the first group children had recent onset of symptoms (maximum 5 days) such as vomiting, diarrhea, respiratory symptoms and one had chest pain. The second group was composed by children with chronic symptoms like chronic abdominal pain and constipation and intermitent tachypnea. The last group was represented by asymptomatic children [8].

Plain chest x-ray can confirm the CDH, as the gastric bubble or intestine loops can be seen in the thorax [1], but a normal chest x-ray does not exclude a CDH, as we could see in the 14 year-old female who had a normal chest x-ray one year before the diagnosis of CDH. Further imaging studies might be needed to confirm the diagnosis and characterize the hernia contents and bowel perfusion.

It is well known that most of the children with CDH associate other anomalies, these being present in 10 to 50% of the cases [1]. Even though the children with late presenting CDH might have been asymptomatic or oligosymptomatic they should undergo further investigations to identify associated anomalies.

Anterior diaphragmatic defect, causing Morgagni hernia, was present in 3 out of 5 patients in our study. Some authors report that the anterior diaphragmatic defect represents less than 2% of the diaphragmatic defects [1] while others claim that the anterior diaphragmatic defect is being present in 25-30% of the diaphragmatic defects [9]. Moreover, Morgagni hernia is reported to be more frequently discovered at older ages.

In both patients with posterolateral diaphragmatic defect, this was present on the left side. The diaphragmatic defect is present on the left in 90% of the cases, while only 10% of the patients with CDH have a right diaphragmatic defect [10-12].

All the children with late presenting CDH must undergo surgical repair, either by classical approach or by minimally invasive surgery (thoracoscopic, laparoscopic) and even though a favorable outcome is usually expected, one should always take into consideration that postoperative complications can appear, such as the case of 14 year-old female patient in our study.

Conclusions

Late presenting CDH is uncommon, but not rare and even though its' diagnosis is difficult, due to nonspecific gastrointestinal and respiratory symptoms, it is essential to be made rapidly in order to prevent certain complications such as stomach or bowel strangulation and cardiac arrest.

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