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Case Report

Diagnostic challenges in late-presentation congenital diaphragmatic hernia: A case study of a 10-month-old with respiratory symptoms ☆,☆☆

Mohammad G. Ibdah[a,*], *Majd Issa*[b], *Suliman N. Berawi*[c], *Nafe' Abu Alwan*[c], *Abdelrahman Abosleem*[a], *Ward Basalat*[d], *Ola Jawabreh*[c], *Layth Al-Karaja*[a]

[a] *Faculty of Medicine, Al-Quds University, Jerusalem, Palestine*
[b] *Faculty of Medicine, Tula State University, Tula, Russia*
[c] *An-Najah National University, Nablus, Palestine*
[d] *Pediatric Surgery Department, Rafidia Governmental Hospital, Nablus, Palestine*

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ABSTRACT

Congenital diaphragmatic hernia (CDH) is a developmental disorder in which the diaphragm, the muscle that separates the chest from the abdomen, does not close during prenatal development, allowing abdominal organs to herniate into the chest cavity. It occurs mainly on the left side (80%-85% of cases). CDH is often identified during prenatal assessment. However, instances of late-presenting CDH beyond infancy are exceedingly uncommon, contributing to frequent misdiagnosis and delayed therapeutic intervention. We present a case of a 10-month-old female with an uneventful antenatal and perinatal history who presented with respiratory distress and multiple episodes of vomiting. Her vital signs were stable upon arrival, but she was sent to the PICU due to hypoactivity, reduced oral intake, and agitation. After an urgent CT scan, a herniation of the small and large bowel loops into the right hemithorax was discovered, along with a defect in the right hemidiaphragm. This resulted in a pleural effusion on the right side, a partially collapsed left lung, and a mediastinal shift to the left. The diaphragmatic hernia was corrected through a lateral thoracotomy at the sixth rib with multiple interrupted sutures, and a chest tube was then inserted into the pleural space above the diaphragm following a smooth reduction of the bowl. This case highlights the importance of early diagnosis, appropriate clinical investigation, and treatment. A good prognosis can be anticipated by promptly discovering and examining the condition.

☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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* Corresponding author.
E-mail address: mribdah@gmail.com (M.G. Ibdah).

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Introduction

Congenital diaphragmatic hernia (CDH) occurs due to a defect in the incomplete muscularization of the diaphragm. Usually, this condition occurs in the neonatal period, presents with respiratory distress symptoms, and could be associated with pulmonary hypoplasia. The late-presenting CDH, however, is relatively infrequent and usually occurs in later childhood, consisting of vague gastrointestinal or respiratory symptoms [1]. The incidence of reported late-presenting CDH varies from 5% to 45.5% of all cases of CDH [2,3]. Diagnosing CDH in the childhood period can be challenging due to the nonspecific broad spectrum of symptoms that may vary from mild to moderate gastroesophageal reflux and respiratory distress that does not respond to medical therapy or as an incidental finding on a routine chest X-ray done for suspected respiratory infection [4]. We are presenting here a case of a 10-month-old female patient with a history of nonspecific respiratory symptoms whose final diagnosis was a right-sided diaphragmatic hernia. This case report highlights the importance of familiarizing with the natural history of this condition in childhood and underscores the course of management compared to neonatal CDH.

Presentation

This 10-month-old girl came to the ER complaining of respiratory distress and vomiting that is not bloody, not gelatinous, and not projectile, with decreased oral intake and suboptimal activity, all of which started today. No trauma, no rash, no diarrhea, no fever, no runny nose, no altered level of consciousness, and no signs of child abuse.

On inspection, the child was hypoactive, stressed, and slightly dehydrated, so he was admitted to the pediatric intensive care unit.

Examination of the child reveals good tone and posture. Auscultation of the chest shows decreased air entry on the right side, without crepitation or wheezing. The abdomen is soft and without HSM or masses.

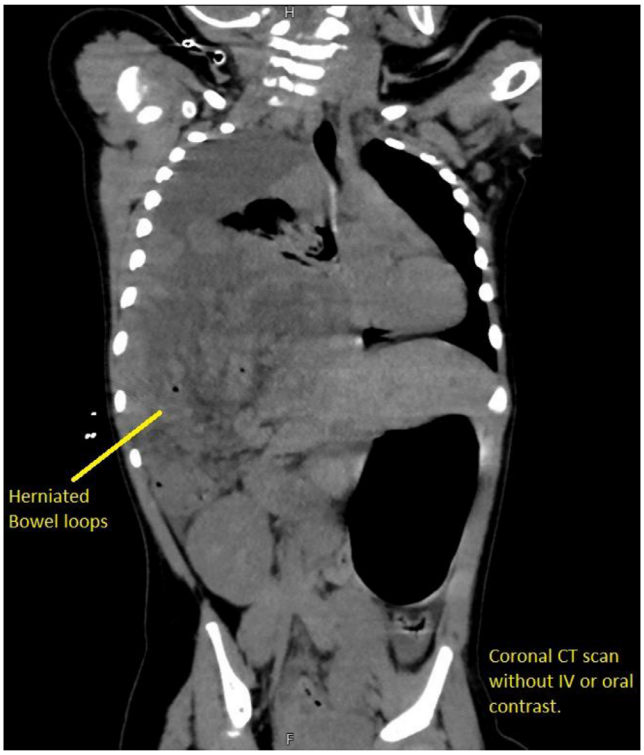

Fig. 1 – Coronal CT shows herniated bowel loops.

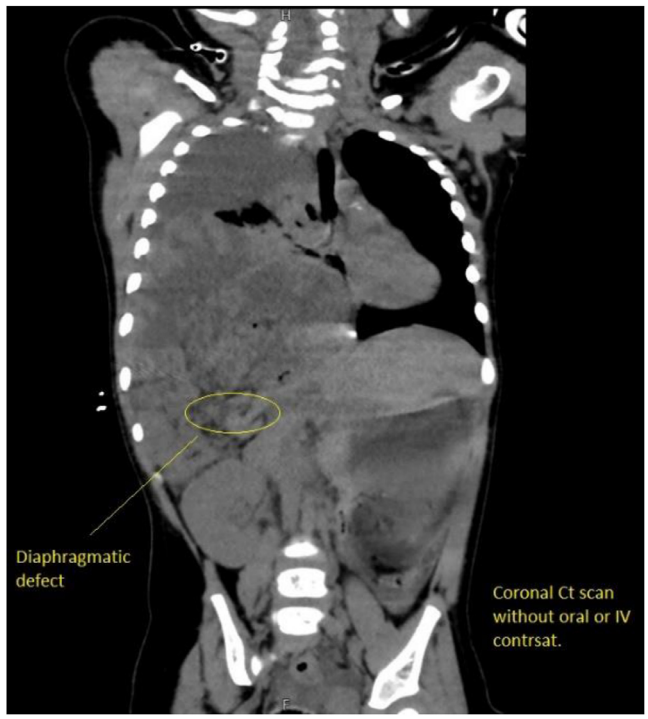

Fig. 2 – Coronal CT shows diaphragmatic defect.

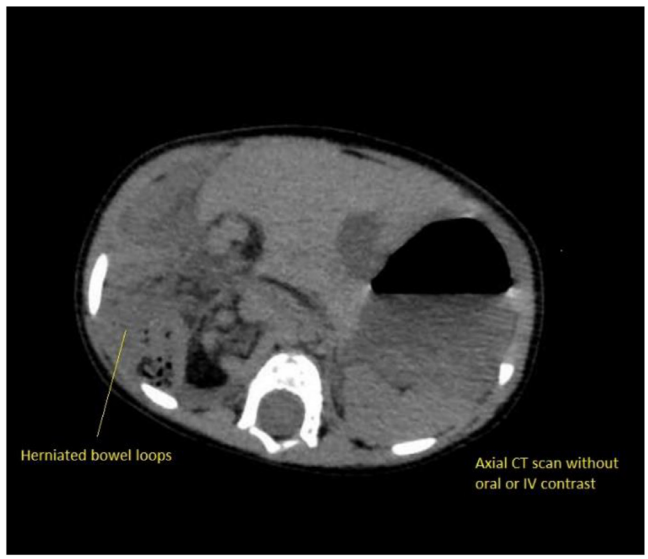

Fig. 3 – Axial CT shows herniated bowel loops.

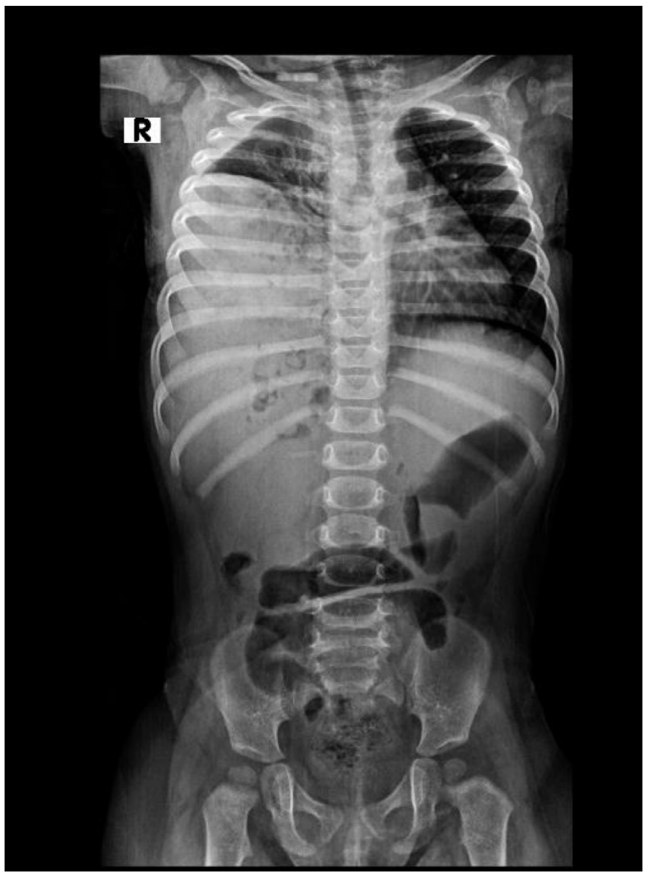

Fig. 4 – Fluid in the right chest and mediastinum shift to the left side.

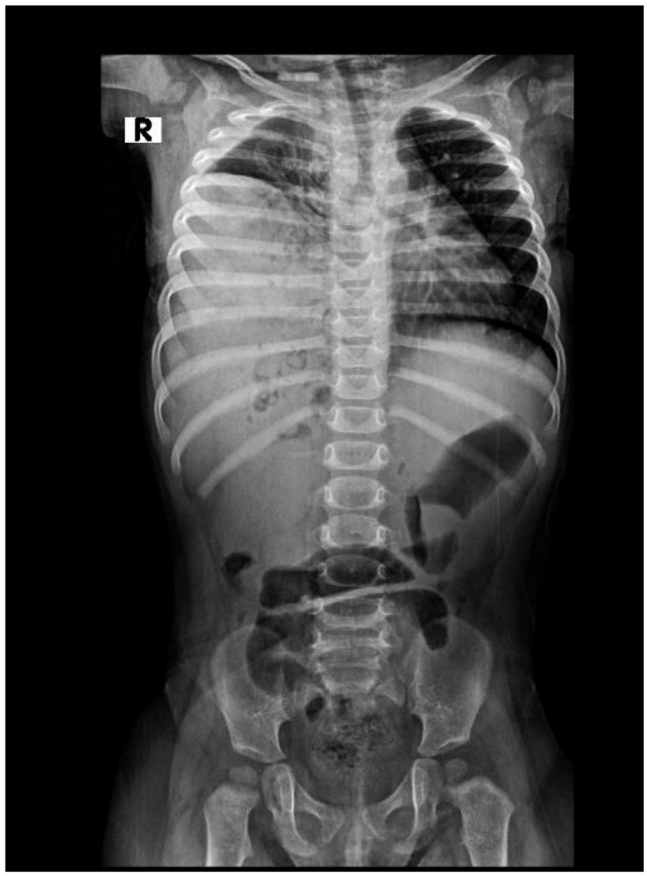

Fig. 5 – Opacification in the right chest and mediastinum shift to the left side.

Vital signs: rectal temperature 36.7, O2 saturation 95% on room air, blood pressure 100/60, heart rate 120, and weight 8.9 kg.

Labs

ABGs before fluid administration: PH = 7.2, H = 12, after resuscitation: PH = 7.3, H = 18.

The lactic acid value was ER 4.5; after the fluid, it was lowered to 2.2.

Abdominal X-ray and chest X-ray (Fig. 4 and 5) were performed and revealed either a diaphragmatic event or a hernia.

The abdominal ultrasound image shows that the liver has been pushed to the left side, mainly through herniated intestinal loops into the right side of the chest. The liver, spleen, gallbladder, and both kidneys are largely normal. The urinary bladder is partially filled, and there is no free fluid.

A CT scan revealed a diaphragmatic hernia with small bowel contents in the right chest (Figs. 1-3). Therefore, the patient was scheduled for an urgent surgical reduction and repair of the hernia, (Figs. 6-8) shows X-rays of the patient chest and abdomen immediately after the surgical repair, postoperative day 1, and before the discharge respectively.

Discussion

This case illustrates the challenges associated with diagnosing late-presenting congenital diaphragmatic hernia (CDH). Congenital diaphragmatic hernia (CDH) is a well-recognized entity occurring in 1 out of 25,000 to 30,000 babies [5]. Incidence is around 1 in every 30,000 live births, male-to-female ratio was approximately 2: 1 [6,3]. The incidence of late diagnosis is variable, ranging from 3% to 5% [7].

It is an acquired herniation of the abdominal viscera that occurs through a congenital defect that the spleen or liver had occluded [8,9]. Beyond the neonatal period is not uncommon.

In our case, a 10-month-old female presented with respiratory distress and vomiting, symptoms that initially led to a broad differential diagnosis. Its diagnosis in infancy and early childhood poses a challenge owing to different clinical presentations ranging from gastrointestinal to respiratory symptoms. These neonates are usually misdiagnosed as having pneumonia until radiological imaging picks up the defect during routine scans for worsening respiratory symptoms [10].

There is a high association of pulmonary hypoplasia and pulmonary hypertension. The degree of lung hypoplasia and pulmonary hypertension are key determinants of the morbidity and mortality of these patients [11]. The commonest 2

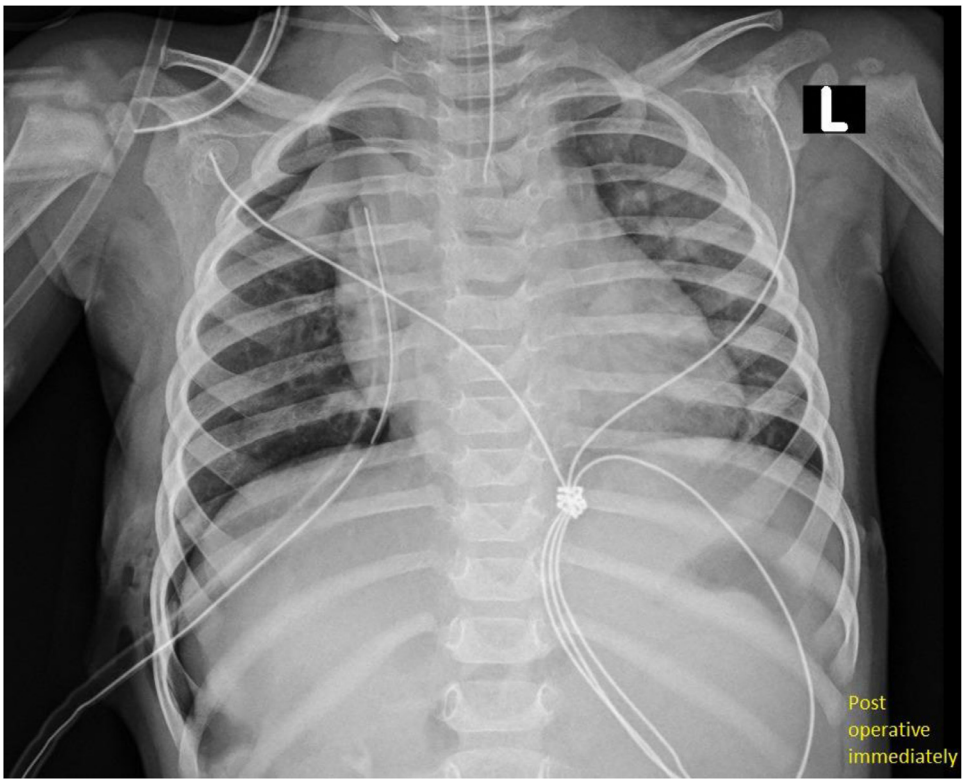

Fig. 6 – Immediate postoperative X-ray.

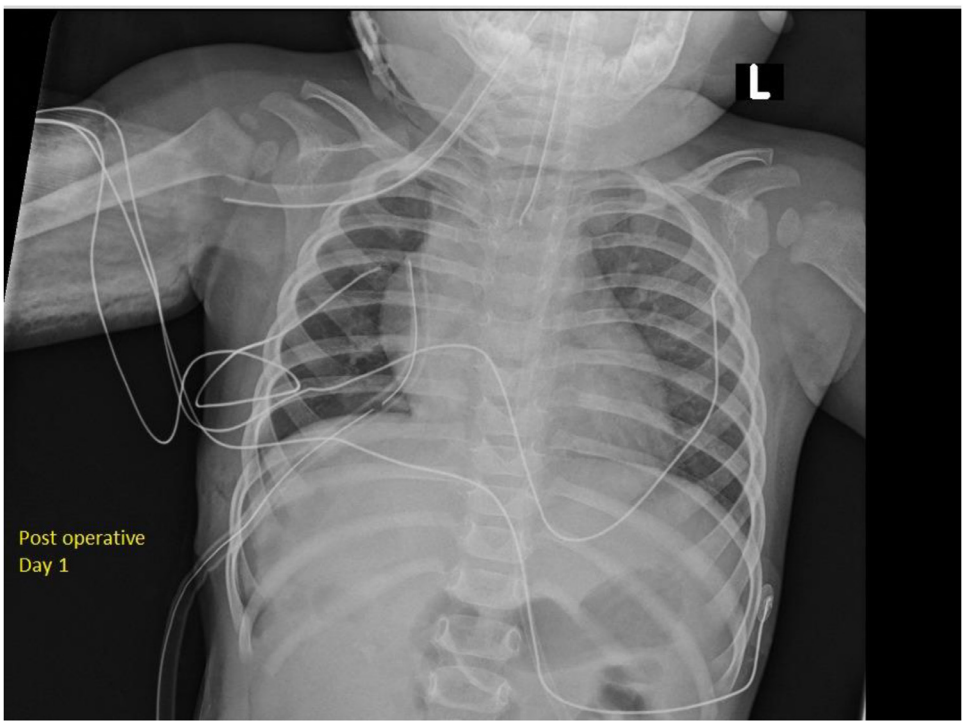

Fig. 7 – Postoperative day 1 X-ray.

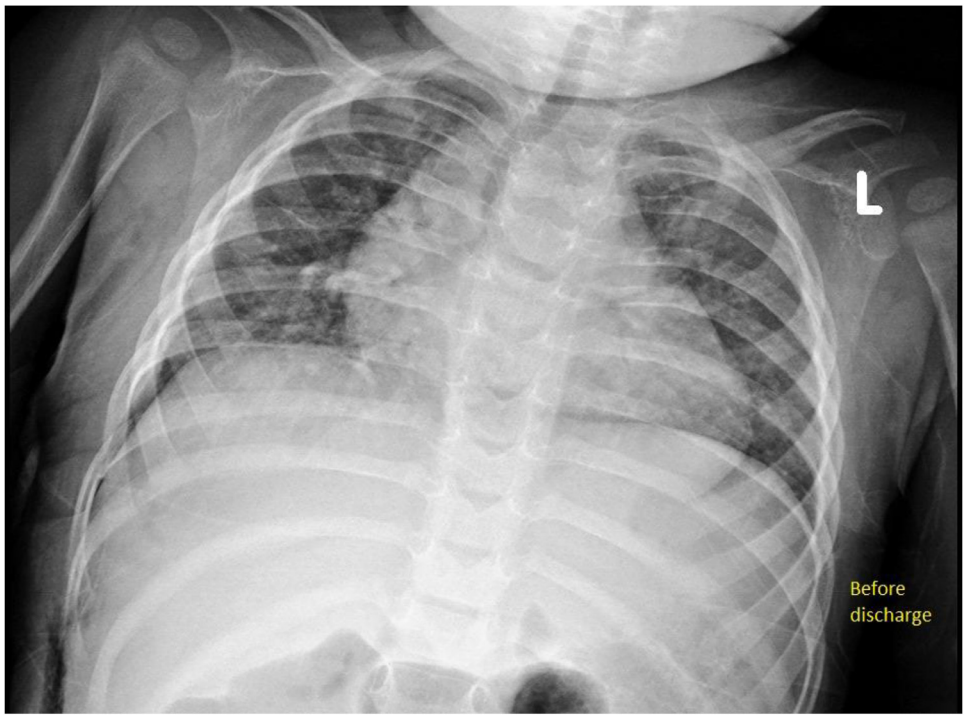

Fig. 8 – X-ray just before patient discharge.

types of foramina where the hernia can occur are Bochdalek and Morgagni. They can be unilateral or bilateral.

The patient's initial evaluation, including a chest X-ray and abdominal ultrasound, suggested a diaphragmatic event or hernia. The diagnosis was confirmed by a CT scan, which revealed herniation of the small bowel into the right hemithorax and a defect in the right hemidiaphragm.

The most common is the Bochdalek type. The Bochdalek type has a posterolateral position, which can result in a mediastinal shift on the contralateral side and pulmonary hypoplasia on the same side of the hernia [12,13].

Symptoms comprise a wide range from mild to moderate gastroesophageal reflux to respiratory distress [4] after the neonatal period the symptoms may be varied like recurrent vomiting or upper respiratory infections [14,15]. The diagnosis occurs in 50–90 % percent with antenatal ultrasonography [16]. Others diagnosed by a chest X-ray, which will show an air-fluid level, in addition to the contrast material administration by a nasogastric tube which will help in the diagnosis [17]. Although, the chest CT scan is the most sensitive diagnostic method for diaphragmatic defects [18].

The surgical approach involved reducing the herniated bowel and repairing the diaphragmatic defect, most patients with late-onset congenital diaphragmatic hernia will treated and do well with a surgical repair [19]. A diaphragmatic hernia may lead to acute gastric volvulus [20], in addition to the risk of gastric strangulation, ischemia, perforation, pancreatitis, peritonitis, shock, and death with a mortality of 80% [20,21].

This case underscores the importance of considering CDH in the differential diagnosis of infants with unexplained respiratory symptoms, as early diagnosis and intervention can significantly improve outcomes.

In conclusion, this case emphasizes the importance of maintaining a high index of suspicion for CDH in infants with atypical respiratory or gastrointestinal symptoms. Early and accurate diagnosis, supported by appropriate imaging, is crucial for timely surgical intervention and favorable outcomes. Future research should focus on improving diagnostic strategies and understanding long-term outcomes in late-presenting CDH cases to enhance clinical management and prognosis.

Conclusion

This instance underscores the diagnostic difficulties and crucial management of late-presenting congenital diaphragmatic hernia (CDH) in a 10-month-old girl with unspecific respiratory symptoms. Although relatively rare, late-onset CDH presents significant diagnostic difficulties due to its varied often nonspecific clinical manifestations that can present as mild gastrointestinal symptoms or respiratory distress mimicking common pediatric conditions such as pneumonia. This case highlights the need for a high index of suspicion for congenital diaphragmatic hernia (CDH) in children with unexplained respiratory problems and the importance of immediate X-ray evaluation for definitive diagnosis.

The successful surgical repair in this patient suggests a favorable prognosis with timely intervention. The need for early detection to prevent potential complications like gastric volvulus, strangulation, and other severe morbidities related to untreated CDH was also emphasized. Physicians should be familiar with the natural history and presentation complexity

of CDH beyond the neonatal period so that prompt diagnosis and treatment can be made. Subsequent research should focus on improving diagnostic approaches and determining long-term outcomes among those suffering from late presenting CDH cases thus further enhancing clinical management and prognosis.

Author contributions

Study concept or design: Mohammad Ibdah.

Writing the manuscript: Mohammad G. Ibdah, Majd Issa, Suliman Berawi, Nafe' Abu Alwan, Abdelrahman Abosleem, Ward Basalat, Ola Jawabreh.

Review & editing the manuscript: Mohammad G. Ibdah, Layth Al-Karaja.

Patient consent

A written informed consent was obtained from the patient's parents for publication of this case report (**Diagnostic Challenges in Late-Presentation Congenital Diaphragmatic Hernia: A Case Study of a 10-Month-Old with Respiratory Symptoms**) and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Ethical approval

Informed consent was signed by the patient's parents for publication.

Provenance and peer review

Not commissioned, externally peer-reviewed.

REFERENCES

[1] Al Ghafri M, Al Sidairi I, Nayar M. Late presentation of congenital diaphragmatic hernia: a case report. Oman Med J 2014;29(3):232.

[2] Elhalaby EA, Abo Sikeena MH. Delayed presentation of congenital diaphragmatic hernia. Pediatr Surg Int 2002;18:480–5.

[3] Bagłaj M. Late-presenting congenital diaphragmatic hernia in children: a clinical spectrum. Pediatr Surg Int 2004;20:658–69.

[4] Hamid R, Baba A, Shera A, Wani S, Altaf T, Kant MH. Late-presenting congenital diaphragmatic hernia. Afr J Paediatr Surg 2014;11(2):119–23.

[5] Malekzadegan A, Sargazi A. Congenital diaphragmatic hernia with delayed presentation. Case Rep Surg 2016;2016:7284914.

[6] Chandrasekharan PK, Rawat M, Madappa R, Rothstein DH. Congenital diaphragmatic hernia: a review. Matern Health Neonatol Perinat 2017;3:1–16.

[7] Kim DJ, Chung JH. Late-presenting congenital diaphragmatic hernia in children: the experience of single Institution in Korea. Yonsei Med J 2013;54(5):1143–8.

[8] Newman BM, Afshani E, Karp MP, Jewett TC Jr, Cooney DR. Presentation of congenital diaphragmatic hernia past the neonatal period. Arch Surg 1986;121(7):813–16.

[9] Wiseman NE, MacPherson RI. Acquired congenital diaphragmatic hernia. J Pediatr Surg 1977;12:657–65.

[10] Mwamanenge NA, Mussa F, Nyamuryekung'e MK, Mkony M, Abdallah Y, Manji K. Late diagnosis of congenital diaphragmatic hernia: a case report. J Med Case Rep 2023;17(1):240.

[11] Canadian Congenital Diaphragmatic Hernia CollaborativeDiagnosis and management of congenital diaphragmatic hernia: a clinical practice guideline. CMAJ 2018;190(4):103–12.

[12] Chang S, Lee H, Yeung C, Chan W, Hsu C, Kao H, et al. A Twenty-year review of early and late-presenting congenital Bochdalek diaphragmatic hernia: are they different clinical spectra? Pediatr Neonatol 2010;51(1):26–30.

[13] Gupta AP, Fouad D, Kays D, Quintana J, Kucera JN. Diaphragmatic hernia: what the radiologist needs to know. Appl Radiol 2020;49:34.

[14] Zaleska-Dorobisz U, Bagłaj M, Sokołowska B, Ładogórska J, Moroń K. Late presenting diaphragmatic hernia: clinical and diagnostic aspects. Med Sci Mon Int Med J Exp Clin Res 2007;13(Suppl 1):137–46.

[15] Berman L, Stringer D, Ein SH, Shandling B. The late-presenting pediatric Bochdalek hernia: a 20-year review. J Pediatr Surg 1988;23(8):735–9.

[16] Enrica B, Paola M, Stefania DV, Elena P, Samanta T, Isabella G, et al. Congenital asymptomatic diaphragmatic hernias in adults: a case series. J Med Case Rep 2013;7:125.

[17] WuCT Huang JL, Hsia SH, Lin JJ, Lai SH. Late-presenting congenital diaphragmatic hernia in pediatric emergency room: two case reports. Eur J Pediatr 2009;168(8):1013–15.

[18] Planting NS, Hoffman-Haringsma AK. [Late presentation of congenital diaphragmatic hernia]. Ned Tijdschr Geneeskd 2017;161:D960.

[19] Joel AF, John L, Samuel E, James F, Louis MB. Diaphragmatic hernia masquerading as pneumothorax in two toddlers case report. Ann Emerg Med 1993;22:1221–4.

[20] Ayala JA, Naik-Mathuria B, Olutoye OO. Delayed presentation of congenital diaphragmatic hernia manifesting as combined-type acute gastric volvulus: a case report and review of the literature. J Pediatr Surg 2008;43:E35–9.

[21] Vandy FC, Landrum JE, Gerig NR, Prahlow JA. Death Due to late-presenting congenital diaphragmatic hernia in a 2-year-old child. Am J Forensic Med Pathol 2008;29:75–9.