International Surgery Journal
Bora R et al. Int Surg J. 2026 Jan;13(1):155-157
http://www.ijsurgery.com
pISSN 2349-3305 | eISSN 2349-2902

DOI: https://dx.doi.org/10.18203/2349-2902.isj20254346

Case Report

Beyond the neonatal period: unmasking late presenting congenital diaphragmatic hernia: a case report

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Received: 04 November 2025
Accepted: 09 December 2025

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ABSTRACT

Congenital diaphragmatic hernia (CDH) is a common neonatal emergency characterized by a defect in the diaphragm causing abdominal organs to herniate into the thoracic cavity. Most cases of CDH present in the neonatal period with severe respiratory distress; however, a proportion of CDH cases present beyond the neonatal period, termed as late-presenting CDH. Being relatively uncommon compared to neonatal CDH, late-presenting cases pose unique diagnostic and therapeutic challenges. In this case report we present an uncommon case of late presenting CDH where posterior lip of diaphragm was completely absent with underdeveloped anterior lip, managed successfully using local tissues and intricate surgical manoeuvres lead to favourable outcome.

Keywords: Neonate, Congenital diaphragmatic hernia, Large defect, Toldt's fascia

INTRODUCTION

Congenital diaphragmatic hernia (CDH) is a common neonatal emergency characterized by a defect in the diaphragm allowing abdominal organs to herniate into the thoracic cavity. Most cases of CDH presents in the neonatal period with severe respiratory distress due to pulmonary hypoplasia and persistent pulmonary hypertension; a proportion of CDH cases present beyond the neonatal period, termed as late-presenting CDH.[1,2] The reported incidence of late-presenting CDH ranges widely from 5% to 45% of all CDH cases.[1]

Despite being relatively uncommon compared to neonatal CDH, late-presenting cases presents with variable and nonspecific signs and symptoms, leading to frequent diagnostic delays or misdiagnosis but generally carry a more favourable prognosis if diagnosed early.[1,3-5] We report here a 6-month-old child with late presenting CDH with deficient posterior lip of diaphragm successfully managed by using Gerota's fascia (Toldt's fascia).

CASE REPORT

A 6 months old female child born preterm at 33 weeks, asymptomatic at birth presented with recurrent vomiting and fast breathing for about a month. At the time of examination, her respiratory rate is 56 breaths per min, SpO_2 93% at room air and afebrile. There is no history of fever, rashes and abdominal distension. On auscultation of chest bowel sounds are audible on left side, abdomen is scaphoid with subcostal recession. Chest X-ray suggestive of left sided CDH with bowel loops visible in left hemi thorax (Figure 1). There was no antenatal anomaly on ultrasonography. Preoperative evaluation with routine blood investigations were normal, however 2D Echocardiography revealed mild to moderate pulmonary hypertension. Subsequently she was planned for urgent operation due to severe tachypneoa and chest retractions. Intraoperative findings included a large left posterolateral diaphragmatic defect (6×4 cm) with absent posterior leaflet and significantly underdeveloped anterior lip containing stomach, left lobe of liver, spleen, small bowel, large bowel and splenic flexure of colon as content along

with complete mal-rotation of midgut. During repair of left congenital diaphragmatic hernia, we made the posterior lip using Gerota's fascia of left kidney (Toldt's fascia) and took deep bites using interrupted non-absorbable sutures through whatever anterior lip present in remnant diaphragm with ICD insertion.

In immediate post-operative period, she was kept on mechanical ventilator for 48 hours and then kept on CPAP for another 72 hours. Chest X-ray in immediate post-operative period showed better lung expansion with residual pneumothorax. She was allowed nasogastric feeds from post-operative day 4 and gradually increased. She tolerated her feeds well and passing urine and stools comfortably. ICD was later removed on POD 8 with serous contents throughout the period with minimal output. Chest X-ray showed better lung expansion (Figure 2). Child discharged on 10th post-operative day and followed up at 1, 3 and 6 month showed good outcome with no recurrence.

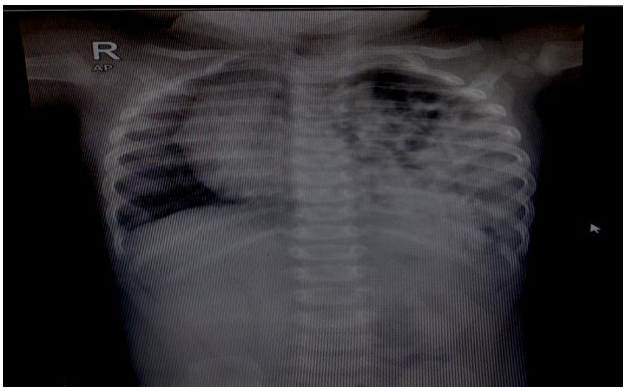

Figure 1: Chest X-ray shows intestinal loops in left thoracic cavity and no stomach bubble, suggesting left sided congenital diaphragmatic hernia.

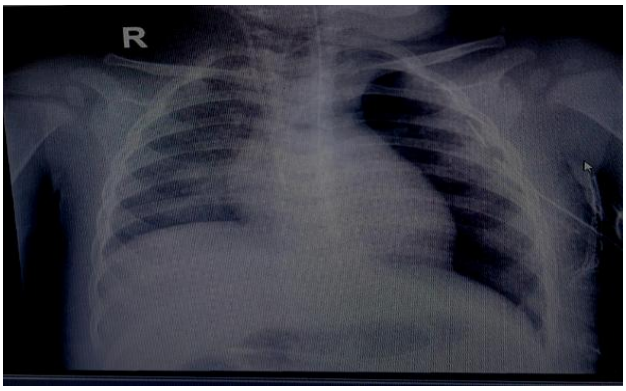

Figure 2: Post- operative chest X-ray showing left lung expansion with ICD in situ.

DISCUSSION

The diaphragmatic defect in late-presenting CDH is identical to that seen in neonates, most commonly involving a posterolateral defect (Bochdalek hernia), although anterior (Morgagni) or central defects can also occur.[6] Timing of herniation plays a pivotal role in clinical outcomes. Classification systems divide CDH into types based on the timing of herniation and resultant pulmonary development.[7] Type 1 and 2 involve early in-utero herniation leading to significant bilateral or unilateral pulmonary hypoplasia and high neonatal mortality. Type 3 refers to herniation occurring later in gestation, where pulmonary hypoplasia is minimal. Type 4 includes herniation occurring after the neonatal period, without pulmonary hypoplasia. Some authors propose type 3 into early-presenting and late-presenting subgroups, with the latter potentially having small, initially asymptomatic hernias that progress later in life. Additionally, some type 4 hernias may remain asymptomatic until triggered by increased intra-abdominal pressure.

The clinical features of late-presenting CDH are variable and nonspecific, leading to frequent diagnostic delays or misdiagnosis.[1] Unlike respiratory distress seen in neonatal CDH, late-presenting cases may present with chronic or intermittent respiratory or gastrointestinal symptoms or they may be asymptomatic and detected incidentally.[8,9] A left-sided hernia is more common, and the absence of a gastric bubble on abdominal imaging may suggest the diagnosis. Also male: female ratio in late-presenting cases is higher 2:1 to 6:1 in various studies.[3,6]

Diagnosing late-presenting CDH remains challenging due to overlapping clinical and radiological features with chest X-ray, though often the first investigation, may be misleading. Passage of a nasogastric tube with its tip visualized in the thoracic cavity on imaging can aid in diagnosis.[1] It may also mimic lower lobe pneumonia, pleural effusion, pneumothorax, lung abscess, or diaphragmatic eventration.[8,10] The presence of gas-filled bowel loops in the thorax or displacement of abdominal organs may be visible on plain radiographs, but confirmatory imaging such as ultrasound, computed tomography, or magnetic resonance imaging may be required.

The associated anomalies in late-presenting CDH have been reported in 8% to 80% of cases. Intestinal mal-rotation and mild pulmonary hypoplasia are the most frequently reported associated abnormalities.[1,3] The presence of a well-formed hernia sac has been reported which may play a role in delaying symptom onset by containing the herniated viscera.[11] The prognosis for late-presenting CDH is usually favourable as compared to neonatal CDH, mainly due to absence of significant pulmonary hypoplasia and pulmonary hypertension.[3,4,5] Once diagnosed, the treatment of late-presenting CDH requires prompt surgical repair. In asymptomatic or stable patients, surgery can be scheduled electively; however, in cases of acute obstruction or respiratory distress, emergency surgery is warranted.[1]

The various surgical options available to cover large CDH defect includes synthetic patch, living tissue like reverse

latissimus dorsi muscle flaps, abdominal muscle flap using internal oblique and transversus abdominis muscle.[12-14] Synthetic patch being most commonly used are associated with patch failure, high recurrence rate and skeletal deformities.[15] Whereas other living tissue flaps requires complex surgical procedures which may be impractical in neonates. Toldt's fascia (TF) flap as described by Okazaki et al involves mobilisation of Gerota's fascia to cover large CDH defect with less recurrence rate and good long term postoperative outcome. Similar results were noted by Fukuzawa et al using Gerota's fascia to repair large CDH defect thoracoscopically.[16,17] Early surgical intervention prevents complications such as bowel strangulation, necrosis, or cardiorespiratory compromise. However, few reported mortalities rate ranging from 40% to 60% particularly in cases with delayed diagnosis.[9] Therefore, despite favourable prognosis, early diagnosis and intervention are important for optimal outcomes. In our case we used left Gerota's fascia (Toldt's fascia) to cover the large CDH defect with deficient posterior lip with small remnant of anterior lip with good postoperative outcome.

CONCLUSION

In large CDH defect with deficient posterior or anterior lip we can consider intricate surgical intervention including Gerota's fascia to cover the defect with good results.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Bora R, Bharadwaj A, Jain N. Beyond the neonatal period: unmasking late presenting congenital diaphragmatic hernia: a case report. Int Surg J 2026;13:155-7.