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CASE REPORT

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Congenital diaphragmatic hernia in patient with 1p36 deletion

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Key Clinical Message

This case underscores the atypical presentation of late-onset congenital diaphragmatic hernia in a 9-old with 1p36 deletion syndrome. Recognition of respiratory distress and abdominal symptoms is crucial for intervention.

Abstract

Congenital Diaphragmatic Hernia (CDH) is a condition characterized by the protrusion of abdominal contents into the thoracic cavity due to a defect in the diaphragm. While typically observed in the neonatal period, CDH can present in later life. This case report describes the presentation, diagnosis, and management of a nine-year-old boy with 1p36 deletion syndrome who presented with respiratory distress, abdominal pain, vomiting, and anorexia. The initial diagnosis was tension pneumothorax, and thus the patient underwent chest tube placement. However, a high-resolution CT scan revealed a left hemidiaphragmatic hernia, and the patient eventually underwent an emergency laparotomy due to acute-onset respiratory distress. Intraoperatively, a diagnosis of Bochdalek hernia with gastric perforation was made, and the CDH and gastric perforations were resolved successfully. This case highlights the importance of considering late-presenting CDH as a possible diagnosis in pediatric patients with similar symptoms and the radiological findings suggestive of tension pneumothorax. Early recognition and prompt surgical intervention can lead to successful management of such cases.

KEYWORDS

1p36 deletion syndrome, congenital diaphragmatic hernia, Hemidiaphragmatic hernia, pediatric, tension pneumothorax

1 | INTRODUCTION

Congenital diaphragmatic hernia (CDH) is a structural defect resulting from incomplete fusion of the pleuroperitoneal membrane that forms the diaphragm, leading to the protrusion of peritoneal viscera into the pleural cavity. The incidence of CDH is 4.8/10,000 live births, and it typically presents with respiratory distress immediately or within the first few hours or days of life.[1] Although rare, CDH can also present at an older age, causing symptoms such as recurrent pulmonary infections, dyspnea, wheezing, abdominal pain, failure to thrive, vomiting, diarrhea, and anorexia.[2] Here, we report a case of a 9-year-old boy with 1p36 deletion syndrome who was presented to the emergency department with shortness of breath, multiple episodes of vomiting, abdominal pain, and decreased oral intake. To the best of our knowledge, this is the first reported case of a late-onset Bochdalek diaphragmatic hernia in a child with 1p36 deletion syndrome.[3] This case is reported following the CARE guidelines, and our aim is to raise awareness among physicians about the possibility of this diagnosis in patients with atypical presentations and its potential association with 1p36 deletion syndrome.

2 | CASE HISTORY AND EXAMINATION

A 9-year-old male with distal 1p36 deletion syndrome and attention-deficit/hyperactivity disorder (ADHD) was presented to the emergency room with mild, constant abdominal pain, multiple episodes of nonbilious vomiting, decreased oral intake, and shortness of breath on the last day. He was admitted to a nearby hospital with sudden onset breathlessness and was diagnosed with tension pneumothorax, based on clinical findings of decreased breath sounds on the left side. The patient was then referred to our hospital for further management. The patient is a product of nonconsanguineous marriage who was born via emergency LSCS at 36+ weeks due to preterm premature rupture of the membranes (PPROM). He was hospitalized during his first week of life due to a chest infection, which was treated with intravenous antibiotics. Motor and developmental milestones were 6 months to a year late. Speech was also delayed, and the patient currently speaks incoherently. In addition, the patient has a learning disability; however, social interactions are normal. Owing to delayed milestones, detailed workups including karyotyping and microarray testing were done, which revealed a distal 1p36 deletion. There was no significant past surgical history. Upon arrival at the ER, the patient had a heart rate of 174 bpm, a blood pressure of 102/70 mmHg, a temperature of 37.78°C, and an oxygen saturation of 95% at 0.5 L of oxygen via nasal cannula.

During the examination, the patient was lethargic and in respiratory distress, with decreased air entry and chest movements on the left side and a GCS of 15/15. He had mild ptosis, micrognathia, prominent ears, and left cheek atrophy. Anthropometric measurements revealed a weight of 13 kg (0.4 percentile), a height of 110 cm (0.4 percentile), and a head circumference of 48.5 cm (0.4% percentile). Muscle tone was slightly decreased. Onchomycosis and poor oral hygiene were observed.

3 | DIFFERENTIAL DIAGNOSIS, INVESTIGATION AND MANAGEMENT

He was initially managed by establishing an IV line and administering supplemental oxygen. Baseline labs Table 1, including CBC, C-reactive protein, blood C/S, CHEM 8, ABG, PT/INR, antibodies, and BSR, were requested. The patient received an IV bolus of normal saline, 130 mg of Paracetamol, STAT doses of Meropenem and Vancomycin, and was nebulized with Ipratropium and normal saline. A CXR revealed a large hyperlucency measuring approximately 18 × 10 cm in the left hemithorax with a significant mediastinal shift to the right Figure 1. A

TABLE 1 Initial laboratory results.

Test conducted	Outcome
Hemoglobin	12.7 (12–15.5 g/dL)
Total leukocyte count	15,100 (4500–11,000/mm^3)
Platelets	328,000 (150,000–450,000/mm^3)
Sodium	145 (135–145 mmol/L)
Potassium	3.8 (3.5–5.0 mmol/L)
Chloride	112 (96–106 mmol/L)
Bicarbonate	17 (96–106 mmol/L)
BUN	40 (7–20 mg/dL)
Urea	85.6 (7–20 mg/dL)
Creatinine	0.5 (0.7–1.3 mg/dL)
C-reactive protein	47.87 (<10 mg/L)
APTT	27.8 (25–35 sec)
PT	12.5 (11–13 sec)
INR	1.17 (0.8–1.1)
pH	7.396 (7.35–7.45)
PCO_2	26.1 (35–45 mmHg)
PO_2	79.8 (75–100 mmHg)
SPO_2	95.7 (95%–100%)
HCO_3	15.7 (22–28 mmol/L)
Base excess	−7.4 (−2–+2 mmol/L)

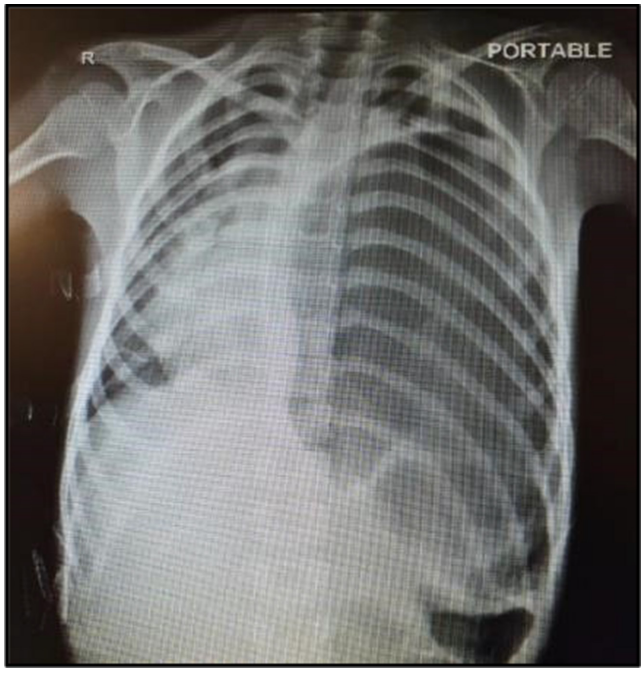

FIGURE 1 Preoperative chest radiograph showing large hyperlucency, and the dimension were approximately 18 × 10 cm in the left hemithorax. A significant mediastinal shift to the right can be observed.

FIGURE 2 Underwater seal connected to the chest tube drain containing 600 mL of brownish aspirate.

needle thoracotomy was done in the left second intercostal space, and bubbles were observed in the underwater seal. Pediatric surgery was enlisted, and a left-sided chest drain was implanted.

The patient was then transferred to the pediatric intensive care unit (PICU) with the diagnosis of tension pneumothorax. His breathing had improved, and he was kept nothing by mouth (NPO). A high-resolution CT scan was carried out, which showed a large left hemidiaphragmatic hernia with predominantly stomach, small bowel loops, and splenic flexure of the colon. The aforementioned remarks resulted in a significant mass effect and contralateral mediastinal shift with a completely collapsed left lung lying posteriorly. A left-sided chest tube with trace hydropneumothorax and a hyperinflated right lung with trace right-sided pleural effusion, were also observed in the scan. On Day 2 of the PICU stay, pediatric surgery was taken on board and a diaphragmatic hernia repair was planned. However, the patient's temperature increased to 38.33°C with sudden onset respiratory distress, tachypnea, tachycardia (180–190 bpm), and restlessness. On examination, the patient had a capillary refill time of 3–4 s. Blood cultures were sent, and the ABGs were taken, and the blood pH was 7.46, the pCO_2 was 29, the pO_2 was 85, the HCO_3 was 20.1, and the lactate was 0.84. The underwater seal of the chest tube drain was filled with 600 mL of brownish aspirate Figure 2.

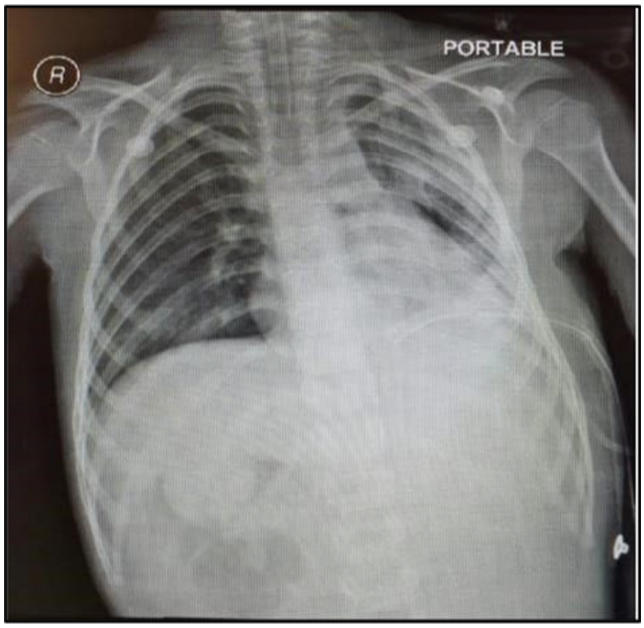

FIGURE 3 Postoperative chest radiograph demonstrates patchy consolidation in the smaller left lung, with a chest tube present on that side obscuring the costophrenic angle. The mediastinum is centrally located, and the right lung appears normal in size.

A plan was devised to electively intubate the patient and pass a nasogastric tube. The patient was shifted to the OR for an emergency laparotomy. Intraoperatively, a left-sided Bockdalek hernia was observed. Stomach, spleen, and bowel loops were seen in the chest, and there was an 8 cm defect in the left diaphragm. The stomach showed perforation in the anterior, with a large collection of brown-colored fluid in the left hemithorax. The bowel

TABLE 2 Reported cases of CDH in the literature.

Reported cases	Patients' relevant demographics	Clinical features	Investigations	Treatment plan
Hyun Beak Shin, Yeon-Jun Jeong[8]	13-year-old boy with no medical history	Abdominal pain and vomiting	Baseline labs, CXR, and CT chest	Emergency laparotomy
Aabha A. Anekar et al.[10]	8-year-old boy with no medical history	Sudden onset dyspnea, chest pain and nonbilious vomiting	Baseline labs, CXR and barium study with an NG tube	Emergency laparotomy
Muhamma d Zahid Abdul Muien et al.[11]	10-year-old boy with no medical history	Postprandial vomiting, epigastric pain, reduced appetite and breathlessness	Baseline labs, CXR and CT thorax	Elective surgical repair

orientation was normal. Gastric perforation and diaphragmatic hernia were repaired, as well as the left chest cavity and peritoneal cavity, which were thoroughly washed. The left chest drain was kept in situ. Postoperatively, the patient was intubated and kept sedated on morphine and midazolam infusions. Amikacin has been added to the list of available antibiotics, joining Vancomycin, Meropenem, and Metronidazole.

4 | CONCLUSION AND RESULTS

CXR, CBC, and CRP were performed post-procedure. For 3 days, the patient was kept NPO. A milrinone infusion was used for persistent tachycardia and poor perfusion. On Day 3, the patient was stable on mechanical ventilation, thus the ventilator settings were reduced. Fever spikes were recorded, and CRP showed a rising trend (44 > 140 > 160). Antibiotics were continued, and the patient was kept NPO. Figure 3 depicts the postoperative chest radiograph.

On Day 4, the patient was successfully extubated and switched to 0.5–1 L of supplemental oxygen via nasal cannula. Total parenteral nutrition was also started. On Day 5, the patient remained vitally stable and was mobilized. Physiotherapy for the chest and incentive spirometry was initiated. CRP decreased from 160 to 83. On Day 6, the patient started taking oral sips. The chest drain, NG tube, and Foley's catheter were removed. CRP continued to decrease (160 > 83 > 23). The patient was transferred from the PICU to the ward on Day 7. He was put on a liquid diet, and his antibiotics were discontinued.

This study demonstrates the importance of considering a late-onset CDH in patients presenting with shortness of breath, abdominal pain, vomiting and reduced appetite. Special care should be taken to correlate radiological findings with clinical symptoms to avoid misdiagnosing and and improper management of CDH. This study explains the diagnosis and management of CDH and sheds light on the association of CDH with chromosomal disorders such a 1p36 deletion syndrome.

5 | DISCUSSION

Late-onset CDH poses diagnostic challenges due to its atypical presentation, often masquerading as respiratory or gastrointestinal issues.[4] The case underscores the importance of considering CDH in the differential diagnosis of patients presenting with non-specific symptoms like recurrent pulmonary infections, abdominal pain, and respiratory distress.[5]

The association between CDH and 1p36 deletion syndrome adds complexity to the genetic landscape of this condition.[4] Understanding the genetic factors contributing to CDH is crucial, as evidenced by its link to single gene mutations, chromosomal abnormalities, and syndromic presentations.[5] The diversity in genetic etiology, including autosomal dominant, autosomal recessive, and X-linked inheritance, highlights the need for a comprehensive genetic evaluation in CDH cases.[6]

Late-presenting CDH, often misclassified as other conditions, necessitates heightened clinical suspicion. The potential for misinterpretation of radiographic imaging emphasizes the need for advanced diagnostic modalities, such as computed tomography or contrast studies, to achieve accurate diagnoses and prevent iatrogenic complications.[7,8]

According to the literature, around 28% of infants with CDH have been reported to have other congenital anomalies.[9] It is important to note that, as with our patient, CDH has been observed in several chromosomal disorders. About one-third of cases have cardiovascular malformations and lesser proportions have skeletal, neural, genitourinary, gastrointestinal, or other defects.[7]

The rarity of late-onset CDH is reflected in limited literature, making each reported case valuable for understanding its clinical spectrum. The case's alignment with previous reports, detailing symptoms like non-bilious vomiting and dyspnea, emphasizes the importance of recognizing these patterns for early diagnosis.[10]

Surgical intervention remains the cornerstone of CDH management, with various approaches available. The

discussion highlights the challenges in choosing the appropriate surgical technique, particularly in emergent cases.[11] The necessity for prompt diagnosis and intervention is evident, as delayed recognition, as seen in this case, can lead to complications such as gastric perforation.[11]

The case's unique aspect involves the emergency laparotomy due to rapid clinical deterioration, emphasizing the need for timely intervention to prevent life-threatening complications.[12,13] The postoperative care, including elective intubation, NG tube insertion, and antibiotic prophylaxis, showcases the multidisciplinary approach required for comprehensive management.

Table 2 summarizes reported cases of congenital diaphragmatic hernia (CDH) in the literature, showcasing diverse patient demographics, clinical presentations, investigative approaches, and treatment plans. These cases contribute valuable insights for clinicians and researchers in managing CDH, emphasizing the need for tailored interventions based on individual characteristics.

AUTHOR CONTRIBUTIONS

Midhat Zihra: Conceptualization; data curation; writing – original draft. **Ibad Rehmaan:** Conceptualization; writing – original draft. **Saman Amjed:** Formal analysis; writing – original draft. **Khawar Abbass:** Methodology; supervision; validation. **Ata ullah Khan:** Project administration; visualization; writing – original draft. **Anwaar ul Haq:** Data curation; formal analysis; software; supervision. **Hashim Talib Hashim:** Project administration; supervision. **Khadija Iqbal:** Investigation; writing – original draft. **Ahmed Dheyaa Al-Obaidi:** Data curation; formal analysis; validation. **Ahmed Qasim Mohammed Alhatemi:** Funding acquisition; investigation; methodology; project administration; writing – original draft; writing – review and editing. **Ali Talib Hashim:** Data curation; formal analysis; supervision; validation.

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CONFLICT OF INTEREST STATEMENT

We declare that we have no conflict of interest.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available from the corresponding author upon reasonable request.

CONSENT

Written informed consent was obtained from the patient's parent to publish this report in accordance with the journal's patient consent policy.

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