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ORIGINAL RESEARCH

Differentiating Imaging Characteristics of Congenital Diaphragmatic Hernia and Diaphragmatic Eventration

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Objective: Congenital diaphragmatic hernia (CDH) and congenital diaphragmatic eventration (CDE) exhibit overlapping imaging features that can contribute to diagnostic challenges. The aim of this study is to systematically and retrospectively examine the clinical and imaging characteristics of CDH and CDE to delineate their differences in diagnosis, treatment, and prognosis assessment, thereby providing a foundation for evidence-based clinical decision-making.

Methods: A retrospective analysis was conducted on 78 cases of CDH and 20 cases of CDE diagnosed between January 2020 and December 2024 at a single institution. Imaging modalities reviewed included chest radiography, gastrointestinal contrast studies, ultrasonography, and computed tomography, in conjunction with clinical data. Key imaging parameters assessed were the integrity of the diaphragmatic contour, diaphragmatic motion, mediastinal displacement, thoracoabdominal organ position, pulmonary development, and amniotic fluid volume.

Results: The sensitivity of X-ray and CT was (CDH 97.43% vs 100%, CDE 80.00% vs 90.00%), and the sensitivity of CT was higher than that of X-ray. The examination results were significantly associated with disease classification, which had statistical significance ($P < 0.05$). In terms of diaphragm integrity and other malformations, CT examination showed significantly higher sensitivity than X-ray examination (100% vs 91.02%,6.41% vs 2.56%). However, in terms of paradoxical movement of the diaphragm, X-ray dynamic examination showed higher sensitivity than CT examination (80.00% vs 0%). Surgical management of CDH was more complex, influenced by the type and extent of the hernia as well as associated anomalies, and often necessitated intensive postoperative care. In contrast, surgical intervention for CDE was less complicated and associated with more favorable outcomes.

Conclusion: Despite certain overlapping imaging findings, CDH and CDE present distinct radiologic and clinical profiles. Comprehensive comparative imaging analysis enhances the understanding of their underlying pathophysiological differences, facilitates accurate diagnosis, and supports the development of tailored management strategies to improve clinical outcomes and long-term quality of life.

Keywords: congenital diaphragmatic hernia, diagnosis, diaphragmatic eventration, imaging, prognosis

Introduction

Congenital diaphragmatic hernia (CDH) is a structural developmental anomaly with Non-cystic lesion characterized by a defect in the diaphragm that permits herniation of abdominal viscera into the thoracic cavity.[1] The estimated incidence is approximately 1 in 2000 live births, representing around 8% of all major congenital malformations. CDH is associated with significant neonatal morbidity and mortality. Currently, no definitive prenatal or early postnatal prognostic markers are available to accurately predict clinical outcomes in patients affected by CDH.[2] Advances in prenatal imaging have facilitated the antenatal detection of approximately 23% of CDH cases, with a reported overall survival rate of 62%.[3]

Congenital diaphragmatic eventration (CDE) is a rare diaphragmatic condition characterized by an abnormal elevation of the diaphragm due to congenital or genetic etiologies, while the anatomical continuity and structural integrity of the diaphragm

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remain preserved. CDE can be classified as partial or complete, with many cases remaining asymptomatic. However, some patients may present with respiratory, gastrointestinal, or cardiovascular symptoms, highlighting the importance of timely diagnosis and accurate assessment.[4] Radiographically, CDE typically presents as diaphragmatic elevation, which may be attributed to diaphragmatic paralysis; paradoxical movement may also be observed via fluoroscopic imaging.[5] Due to limited spatial resolution, conventional radiography often lacks the capability to accurately evaluate diaphragmatic thickness or function. In contrast, ultrasonography offers improved assessment by enabling visualization of diaphragmatic thickness as well as dynamic contraction and motion, particularly in the context of dysfunction.[6] Computed tomography (CT) provides high-resolution imaging of diaphragmatic anatomy, further aiding diagnostic evaluation. Imaging modalities are thus central to the diagnosis, differentiation, and clinical management of both CDH and CDE.

The aim of this study is to enhance diagnostic precision in differentiating CDH and CDE by systematically comparing their respective imaging features. The findings are expected to provide a more reliable evidence base to inform surgical planning and prognostic evaluation, with the ultimate goal of improving health outcomes and quality of life for affected patients.

Information and Methods

Participants of the Study

Through random sampling, imaging and clinical data were collected and analyzed for cases diagnosed between January 2020 and December 2024 at the study institution. A total of 78 cases of CDH and 20 cases of CDE were included. Inclusion criteria: Meet the diagnostic criteria of CDH and CDE; Complete clinical data; age ≤13 years. Exclusion criteria: contraindications of X-ray or CT examination; severe heart, liver and kidney diseases; immune dysfunction or mental disorder patients. The study was approved by Ethics Committee of the Children's Hospital of Shanxi and Women Health Center of Shanxi (Approval No. IRB-WZ-2025-017). Informed consent was taken from all the patients' parents or legal guardians.

Among the 78 CDH cases, all patients were in the fetal, neonatal, or infant stages, with the average age corresponding to the neonatal period. Reported clinical manifestations included shortness of breath (8 cases), respiratory distress (70 cases), vomiting (66 cases), and cyanosis (68 cases). All 78 patients underwent surgical intervention at the institution, and 3 cases resulted in mortality.

In contrast, the 20 cases of CDE involved patients ranging in age from the neonatal period to school age, representing a broader age distribution. Clinical presentations included mild respiratory distress (5 cases), exertional dyspnea (3 cases), and reduced appetite (2 cases), while 15 patients were asymptomatic. All 20 patients were discharged, with no deaths.

Methods

X-ray (Siemens AXIOM Aristos VX): Patients were positioned supine with radiation protection. Standard fluoroscopic parameters were applied to assess diaphragmatic motion. Fluoroscopy (Siemens AXIOM ICONOS R200): 48–52 kV, 2.3–3 mA, 1.2 m SID; oral iohexol 20 mL after 2–3 h fasting; monitoring under 70–100 kVp, 100–500 mA, 0.01–0.1 s exposure. Chest CT (Philips Brilliance 16-slice): pre-medication with chloral hydrate, removal of metal objects, respiratory gating; 100 kV, 70 mA, 1–2 mm slice, 0.5–1 mm interval. Images acquired by a senior technician and reviewed by two associate-senior physicians. Imaging analyses were independently conducted by two experienced physicians using a double-blind method. In instances of diagnostic discrepancy, a consensus was reached through discussion between the two reviewers. The final diagnosis was determined based on a comprehensive evaluation that incorporated both imaging findings and surgical pathology results.

Observation Indicators

Statistical analyses evaluated age distribution, clinical characteristics, imaging features, and surgical/prognostic outcomes between groups.

Statistical Treatment

SPSS20.0 was used to analyze the data, and the count was expressed as n (%). The accuracy, sensitivity and imaging manifestations of CDH and CDE diagnosis by X-ray and CT were analyzed and compared, and the X^2 test was used.

Results

Table 1 shows the average age of CDH was less than 30 days, while the average age of CDE was more than 1 year.

The imaging findings and diagnostic performance are summarized in Table 2. The sensitivity of X-ray and CT as follows: CDH (97.43% vs 100%) and CDE (80.00% vs 90.00%), with CT sensitivity significantly higher than X-ray ($P < 0.05$). The examination results were significantly associated with disease classification, with statistical significance.

Table 3 reveals that CDH was associated with more severe clinical manifestations, including cyanosis, respiratory distress, and gastrointestinal symptoms. These cases were frequently accompanied by other congenital anomalies and

Table 1 Age Characteristics

	CDH (78 Cases)	CDE (20 Cases)
Age		
1h-10d	23	1
11d-30d	26	1
1m-5m	19	2
5m-1y	6	3
1y-2y	3	5
2y-5y	1	8
Average Age	<30d	>1y

Table 2 Comparison of X-Ray and CT Examinations for the Diagnosis of CDH (78 Cases) and CDE (20 Cases). Comparison of X-Ray and CT Examinations in the Detection of Diaphragmatic Hernia and Diaphragmatic Elevation (A); Accuracy Analysis of X-Ray and CT Examinations (B)

(A) X-ray and CT showed the case of diaphragmatic hernia and diaphragmatic elevation		
	CDH (78 Cases)	**CDE (20 Cases)**
Clinical diagnosis +	78	20
—	0	0
X-ray examination +	76 (97.43%)	16 (80.00%)
—	2 (2.56%)	4 (20.00%)
X^2	≈8.30	
P	≈0.004	
CT examination +	78 (100%)	18 (90.00%)
—	0 (0%)	2 (10.00%)
X^2	≈7.90	
P	≈0.005	
(B) Comparison of diagnostic value of X-ray and CT in CDH and CDE		
	CDH (78 cases)	**CDE (20 cases)**
X-ray examination		
Percent of accuracy %	100%	100%
Response rate %	97.43%	80.00%
CT examination		
Percent of accuracy %	100%	100%
Response rate %	100%	90%

Notes: The sensitivity of X-ray and CT was (CDH 97.43% vs 100%, CDE 80.00% vs 90.00%), and the sensitivity of CT was higher than that of X-ray ($P < 0.05$). The examination results were significantly associated with disease classification, which had statistical significance.

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Table 3 Clinical Characteristics

		CDH (78 Cases)	CDE (20 Cases)
Symptoms	Breathing difficulty	70	5
	Shortness of breath	8	3
	Cyanosis	68	0
	Vomiting	66	2
	Intestinal obstruction	3	0
	Asymptomatic	2	15
Prenatal diagnosis		2	0
Associated Malformations	Rectal and anal malformations	0	0
	Congenital heart disease	4	1
	Lung hypoplasia	6	0
	Lung sequestration	2	0
	Polydactyly	1	1
	Gastric volvulus	2	6
Surgical treatment		78	8
Discharge		75	8
Death		3	0
No Surgery		0	12

generally required surgical intervention. Conversely, CDE presented with milder symptoms, was infrequently associated with additional malformations, and in most cases did not necessitate surgical treatment.

Table 4 shows that X-ray (gastrointestinal radiography) examination identified mediastinal shift (87.17%), diaphragmatic absence (91.02%), and small intestine hernia (98.58%)—three conditions accounting for a significant proportion. The combination of X-ray and gastrointestinal radiography exhibits high sensitivity but poor specificity in detecting other associated anomalies (0%). Table 4 indicates that CT examination uncovered diaphragmatic absence (100%), thoracic hernia of the small intestine (93.58%), pulmonary hypoplasia (97.43%), and other associated anomalies (6.41%). Sensitivity comparisons between X-ray and CT were (CDH 97.43% vs 100%, CDE 80.00% vs 90.00%), with CT sensitivity superior to X-ray ($P < 0.05$), and these results correlated significantly with disease classification. For

Table 4 Comparison of CDH and CDE Imaging, Respectively in X-Ray (A), CT (B), and Ultrasound (C)

(A) X-ray (Gastrointestinal Contrast) Examination Comparison			
		CDH (78 cases)	**CDE (20 cases)**
Mediastinal shift		68 (87.17%)	2 (10.00%)
Diaphragmatic contour	Diaphragmatic defect or absence	71 (91.02%)	0 (0%)
	Elevated diaphragm, intact	1 (1.28%)	20 (100%)
	Diaphragmatic paradoxical movement	1 (1.28%)	16 (80.00%)
Hernia content in chest	Stomach	21 (26.92%)	0 (0%)
	Small intestine	73 (93.58%)	0 (0%)
	Small intestine and colon	56 (71.79%)	0 (0%)
	Stomach, small intestine, colon	6 (7.69%)	0 (0%)
Lung compression and decreased lung field transparency		78 (100%)	0 (0%)
Misdiagnosis		2 (2.56%)	1 (0.05%)

(*Continued*)

Table 4 (Continued).

(B) CT Examination Comparison			
		CDH (78 cases)	**CDE (20 cases)**
Elevated diaphragm, intact		0 (0%)	20 (100%)
Diaphragmatic defect or absence		78 (100%)	0 (0%)
Hernia content in chest	Stomach	21 (26.92%)	0 (0%)
	Small intestine	73 (93.58%)	0 (0%)
	Small intestine and colon	56 (71.79%)	0 (0%)
	Stomach, small intestine, colon	6 (7.69%)	0 (0%)
Lung compression, underdevelopment		76 (97.43%)	2 (10.00%)
Associated malformations		5 (6.41%)	0 (0%)
(C) Prenatal Ultrasound Comparison			
		CDH (2 cases)	**CDE (0 cases)**
Hernia content in chest		2	0
Mediastinal shift of heart		2	0
Polyhydramnios		1	0

diaphragm integrity and other malformations, CT sensitivity (100% and 6.41%) was significantly higher than X-ray (91.02% and 2.56%). Conversely, X-ray dynamic examination outperformed CT in detecting diaphragmatic paradoxical movement (80.00% vs 0%). X-ray had a higher CDH misdiagnosis rate than CT (2.56% vs 0%), with a slightly higher rate for CDE as well (0.05% vs 0%). In two CDH misdiagnoses, X-ray showed only a blurred, elevated diaphragm with reduced lung transparency, resulting in misclassification as pneumonia or diaphragmatic elevation, later confirmed by CT. One CDE misdiagnosis involved X-ray findings of a blurred, minimally elevated diaphragm—initially diagnosed as pneumonia but later confirmed as CDE via CT. Table 4 shows that two cases were identified early via prenatal ultrasound screening, underscoring its value in early diagnosis. Congenital diaphragmatic elevation primarily manifests as abnormal diaphragmatic elevation. Dynamic chest imaging demonstrates paradoxical movement: the affected diaphragm is significantly elevated (by >2 intercostal spaces), with the healthy side descending and the affected side rising during inspiration. X-ray fluoroscopy highlights complete diaphragmatic elevation (100%) and paradoxical movement (80.00%) as key features, while CT identifies diaphragmatic thinning, elevation, and completeness (100%), plus pulmonary hypoplasia (10.00%).

Table 5 demonstrates that surgical management of CDH was more complex, with prognostic outcomes varying based on disease severity and comorbidities. By comparison, surgical procedures for CDE were relatively straightforward and typically associated with favorable prognoses. Diaphragmatic elevation surgery primarily aims to fold and repair the lax and weak diaphragm, thereby enhancing its tension and restoring normal morphology and function. Unlike compared

Table 5 Surgical and Prognostic Analysis

Item	**CDH (78 Cases)**	**CDE (20 Cases)**
Surgical indications	Almost always requires surgery	Symptoms obvious or ineffective with conservative treatment
Timing of surgery	Neonatal or infant period	Decided based on symptoms
Surgical method	Hernia content reduction + diaphragm repair	Diaphragm folding + fixation
Prognosis	Dependent on lung development and associated malformations	Good prognosis

with complex thoracoabdominal surgeries, this procedure does not involve the removal or reconstruction of vital organs, featuring clearly defined surgical steps and standardized procedures. Surgeons can perform the operation with clear visual guidance through laparoscopy, enabling precise manipulation. Moreover, this technique is well-established, offering shorter operation times and reduced infection risks. That said, individualized treatment strategies were deemed essential for both conditions, tailored to each patient's specific clinical presentation and overall health status.

Discussion

Comparative Analysis of Results

(1) Comparative analysis of clinical characteristics: CDH is typically diagnosed earlier, with some cases identified prenatally via routine fetal screening. Its clinical presentation is often complex, involving respiratory and gastrointestinal symptoms, and may include other congenital anomalies. These factors increase surgical complexity and result in a non-negligible mortality rate. Prior studies indicate that CDH's high morbidity and mortality primarily stem from severe underlying cardiopulmonary dysfunction.[7] In contrast, CDE has milder clinical manifestations and affects a broader age range, with generally favorable surgical outcomes and low mortality. However, recent advances in prenatal diagnostic imaging have enabled earlier detection of both conditions, weakening age as a distinguishing factor between them.

(2) Comparative analysis of imaging characteristics: The sensitivity of X-ray and CT was (CDH 97.43% vs 100%, CDE 80.00% vs 90.00%), and the sensitivity of CT was higher than that of X-ray. The examination results were significantly associated with disease classification, which had statistical significance ($P < 0.05$). In terms of diaphragm integrity and other malformations, CT examination showed significantly higher sensitivity than X-ray examination (100% vs 91.02%, 6.41% vs 2.56%). However, in terms of paradoxical movement of the diaphragm, X-ray dynamic examination showed higher sensitivity than CT examination (80.00% vs 0%). CDH is commonly characterized by a diaphragmatic defect or absence on the affected side, with intestinal loops herniating into the thoracic cavity, mediastinal shift, reduced ipsilateral lung volume, and decreased lung field radiolucency. Excessive amniotic fluid suggests gastrointestinal malformation, requiring further evaluation of fetal lung volume and abdominal organ position. Studies indicate a relationship between excessive amniotic fluid and CDH, but it is not absolute; diagnosis should be confirmed with additional imaging. Chest radiography typically shows air-filled bowel loops in the thorax and diminished abdominal gas (Figures 1 and 2). CT clearly visualizes herniated abdominal organs and their anatomical relationships with thoracic structures (Figure 3). Prenatal ultrasound reveals gastric bubble and intestinal herniation into the thorax, with heart/mediastinum shifted contralaterally and compressed, underdeveloped lung tissue. In contrast, CDE is marked by an abnormally elevated but intact diaphragm. Diaphragmatic eventration is incidental, and fluoroscopy distinguishes it (no paradoxical movement) from paralysis (with paradoxical movement). The diaphragm elevates ≥ 2 intercostal spaces, rising abnormally during inspiration while the contralateral side descends. CT shows a thin, elevated, intact diaphragm without herniation (Figures 4 and 5). Ultrasound enables dynamic assessment, often revealing paradoxical motion. These distinct imaging findings are key to accurate clinical differential diagnosis.

(3) Comparative analysis of surgical and prognostic outcomes: Surgical intervention remains the primary treatment for CDH, with early detection and timely correction key to improving outcomes. Most surgeries are performed during the neonatal or early infancy period. For individuals with severe diaphragmatic defects or associated malformations, prompt surgery is essential; elective procedures may suit those with milder symptoms. Prognosis depends on multiple factors: diaphragmatic defect size, extent of herniated abdominal contents, pulmonary hypoplasia severity, concomitant anomalies, and postoperative care quality. Severe cases may involve complications or mortality. Conversely, CDE with mild or no symptoms may be managed conservatively via observation and routine follow-up. Surgery is considered only for significant symptoms or complications, and CDE's overall surgical prognosis is favorable.

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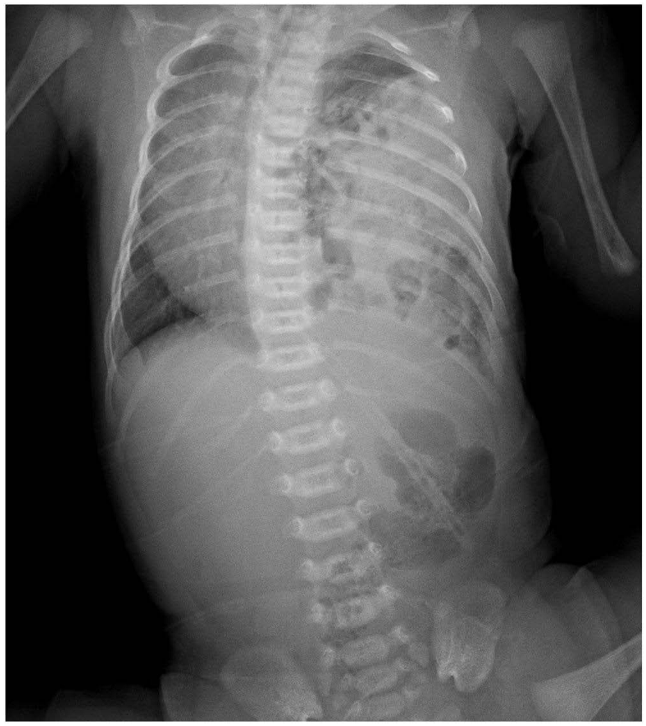

Figure 1 The disappearance of the left diaphragm shadow, more intestinal loops hernia in the left thoracic cavity, and the right displacement of the trachea and mediastinum.

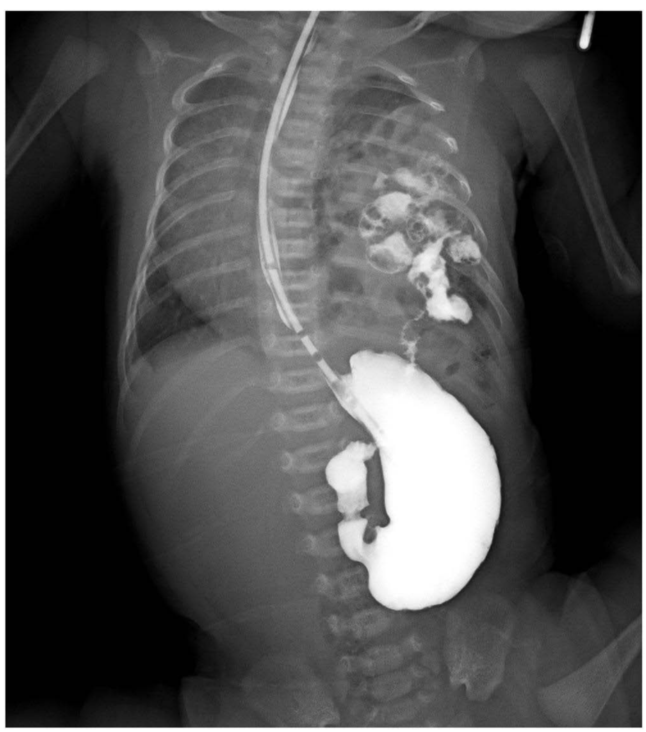

Figure 2 A visible imaging of a bowel loop hernia in the left thoracic cavity and the stomach located in the upper left abdomen.

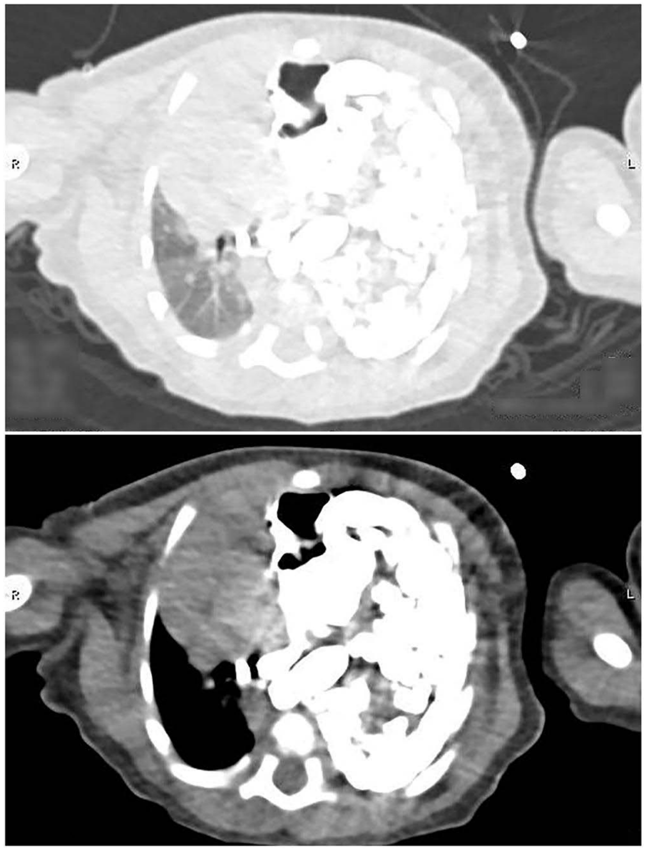

Figure 3 A discontinuity of the diaphragm on the left side, and a hernia of the radiolabeled bowel loop can be seen in the left thoracic cavity.

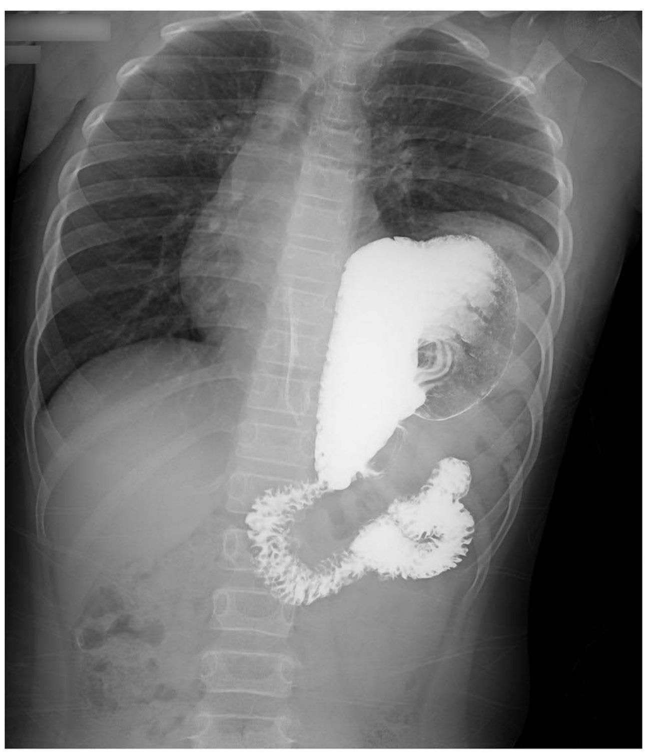

Figure 4 The elevation of the left diaphragm, smooth and continuous, with paradoxical respiratory movement. The gastric bubble and intestinal tube shadows are visible under the left diaphragm.

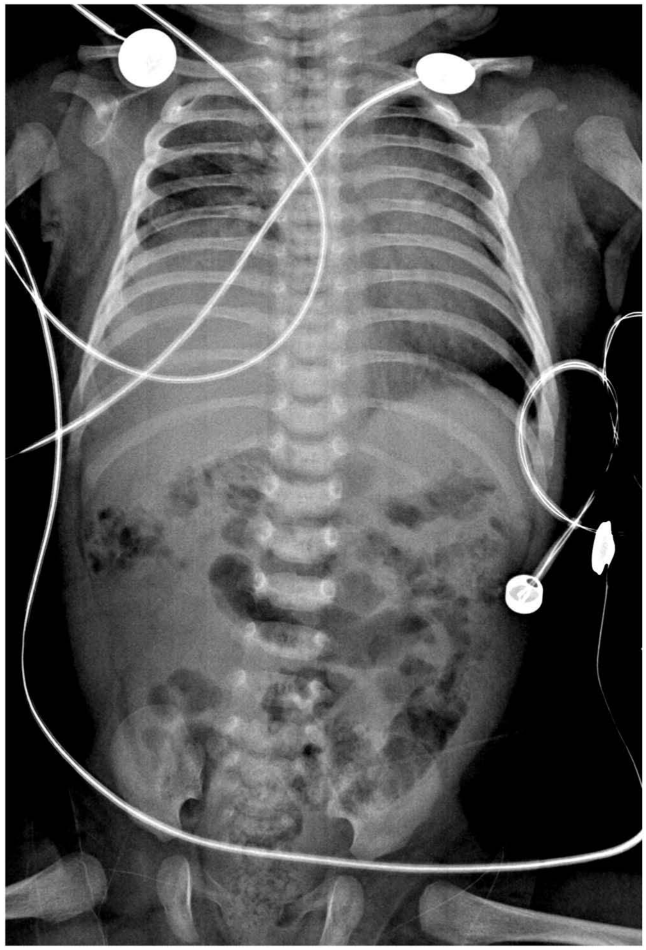

Figure 5 The right diaphragm is elevated by 3 intercostal spaces compared with the left diaphragm, and the diaphragm is smooth and continuous.

Differential Diagnosis of CDH and CDE

This study highlights that rational, effective use of imaging is critical for differentiating CDH from CDE. Past studies, limited by single examination techniques, often led to misdiagnosis or missed cases. Below is an analysis of misdiagnosed cases from our hospital. In this study, two CDH cases were initially misdiagnosed as CDE on plain radiographs due to subtle diaphragmatic defects and no overt herniation. Additionally, one CDE case was misdiagnosed as CDH due to mediastinal shift and indistinct diaphragm caused by abdominal organ compression. Research findings include: (1) In CDE cases with severe abdominal organ compression, the elevated diaphragm may appear indistinct, and associated pulmonary hypoplasia can mimic abdominal organ herniation into the thorax, potentially misdiagnosing CDE as CDH. (2) Small diaphragmatic hernias may show only subtle defects, with minimal/no herniation or mediastinal shift, increasing misdiagnosis as CDE. In summary, typical imaging differences are: congenital diaphragmatic hernia involves more severe lung compression and mediastinal displacement than diaphragmatic elevation, while diaphragmatic elevation rarely causes mediastinal displacement. It manifests as reduced activity and paradoxical movement of the affected diaphragm.

Given these diagnostic challenges, a multimodal imaging approach is essential to comprehensively assess diaphragmatic integrity, mobility, and the anatomical positioning of abdominal organs.

CDH and CDE differ significantly in pathogenesis, imaging, and clinical presentation, though overlapping features may exist. CDH is a relatively rare congenital anomaly involving diaphragmatic developmental defects, characterized by abdominal contents herniating into the thorax. This causes varying degrees of pulmonary hypoplasia and hypertension.[8] In contrast, CDE stems from aberrant muscular development of the embryonic septum transversum (8th–10th weeks of gestation), often with deficient muscle or collagen fibers. This forms a fibrous membrane that allows abnormal bulging of abdominal organs into the thorax.[9]

The hallmark imaging feature of CDE is dome-shaped diaphragmatic elevation, typically composed mainly of fibrous tissue with little to no muscle fibers. Additionally, incomplete diaphragmatic development or absent phrenic nerve innervation contributes to its pathogenesis. Perioperative anesthetic management for CDE patients with ventricular septal defects and severe pulmonary hypertension poses distinct clinical challenges.[10]

CDE often involves paradoxical diaphragmatic motion and typically has mild clinical symptoms, enabling conservative management in most cases. Comparative imaging analysis of CDH and CDE offers valuable insights for early diagnosis, individualized treatment planning, and long-term management, with the primary goal of improving clinical outcomes.

While imaging modalities are key for differentiating CDH from CDE, diagnostic challenges remain. Further research is needed to refine imaging-based differentiation and identify reliable prognostic indicators. Prenatal ultrasound helps families make more informed decisions and reduces their economic and emotional burden by measuring lung-head ratio, fetal lung volume, and liver herniation degree. Advances in prenatal imaging have enabled earlier diagnosis, prenatal interventions, and optimized postnatal management.[11,12] In recent years, integrating artificial intelligence (AI) and machine learning into medical imaging has opened new avenues for improving diaphragmatic eventration diagnosis and management. AI-powered automated imaging analysis has shown potential to enhance assessments of diaphragmatic thickness, mobility, and surrounding organ compression, boosting diagnostic accuracy and treatment efficacy. AI technology analyzes CDH and CDE imaging features to assess diaphragm integrity, positioning, and abdominal organ herniation into the thorax. It aids diagnosis by algorithmically quantifying abnormal indicators. This technology allows doctors to analyze imaging data rapidly, reducing manual review time. Using extensive case data, it detects subtle imaging variations. However, AI has limitations: it heavily relies on high-quality imaging data, and blurry or distorted images may impair diagnostic accuracy. Additionally, its recognition accuracy for complex cases remains suboptimal, making it difficult to integrate with clinical case studies for comprehensive diagnosis. In summary, AI serves as an efficient auxiliary tool but cannot fully replace doctors' integrated judgment.

In conclusion, the accurate differentiation of imaging characteristics and clinical manifestations between CDH and CDE is essential for enhancing diagnostic precision. Future research should further explore the prognostic value of imaging indicators and evaluate AI's clinical application in diagnostic workflows.

Abbreviations

CDH, Congenital Diaphragmatic Hernia; CDE, Congenital diaphragmatic eventration.

Data Sharing Statement

All data generated or analysed during this study are included in this article. Further enquiries can be directed to the corresponding author.

Ethics Approval and Consent to Participate

This study was conducted in accordance with the Declaration of Helsinki and its subsequent amendments. The study was approved by Ethics Committee of Shanxi Children's Hospital, Women Health Center of Shanxi (Approval No. IRB-WZ-2025-017). Informed consent was taken from all the patients' parents or legal guardians.

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Disclosure

The authors declare that they have no competing interests in this work.

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