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Case Report

Prenatal Diagnosis of Congenital Diaphragmatic Eventration: Case Report and Structured Literature Review

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Abstract

Objective: To describe a prenatal case of congenital diaphragmatic eventration, describe prenatal presentation and summarize available postnatal outcomes of previously reported cases. Methods: We present a prenatally diagnosed case of congenital diaphragmatic eventration managed at our institution and a structured literature review to identify previously reported prenatal cases published between 1984 and 2025. Results: Congenital diaphragmatic eventration was diagnosed at 20 + 2 weeks' gestation following suspicion of fetal dextrocardia. Fetal magnetic resonance imaging confirmed diaphragmatic continuity and excluded congenital diaphragmatic hernia. Mild neonatal respiratory distress required only transient continuous positive airway pressure. Nineteen prenatally suspected cases were identified from the literature, most of which were initially misdiagnosed as congenital diaphragmatic hernia (89.5%). Correct prenatal characterization was reported in 57% of cases evaluated with fetal MRI and 25% with ultrasound alone; however, these findings should be interpreted as a descriptive observation due to the limited number and heterogeneity of available reports. Conclusions: Congenital diaphragmatic eventration is a rare but important mimic of congenital diaphragmatic hernia. Fetal magnetic resonance imaging may provide additional anatomical information in selected cases and facilitate counselling and perinatal management.

Keywords: congenital diaphragmatic eventration; prenatal diagnosis; fetal magnetic resonance imaging; congenital diaphragmatic hernia; fetal thoracic anomalies

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1. Introduction

Congenital diaphragmatic eventration (CDE) is a rare diaphragmatic abnormality, accounting for approximately 5% of all congenital diaphragmatic anomalies. It is defined as the abnormal elevation of all or part of an intact but thinned and functionally deficient diaphragm. CDE may be congenital or acquired; the acquired form is typically secondary to reduced diaphragmatic muscle tone, such as in diaphragmatic paralysis caused by

phrenic nerve injury or aging [1]. Historically, the condition was first described by Jean-Louis Petit in 1774, while the term "eventration" was later introduced by Beclard in 1916 [2]. Although not a true hernia, diaphragmatic eventration is characterized by an elevated diaphragm that may compress the developing lung, potentially leading to pulmonary hypoplasia and postnatal respiratory compromise. Although CDE may occur as an isolated anomaly, it has also been associated with chromosomal abnormalities, congenital syndromes, and additional structural anomalies, including congenital heart disease, pulmonary and other systemic abnormalities [3]. Prenatal imaging findings of CDE may closely resemble those of congenital diaphragmatic hernia (CDH) [4], making differential diagnosis challenging. A key diagnostic feature is the visualization of diaphragmatic continuity, which confirms the separation between the abdominal and thoracic cavities. In this context, fetal magnetic resonance imaging (MRI) plays an important complementary role by clearly demonstrating diaphragmatic integrity and the position of abdominal and thoracic organs [5]. Accurate prenatal diagnosis is essential because the prognosis and postnatal management of CDE differ significantly from those of CDH. While mild forms may remain asymptomatic, complete eventration can mimic diaphragmatic hernia with displacement of abdominal organs into the thorax [5,6]. Congenital diaphragmatic anomalies may impair fetal lung development; however, in focal or less severe forms of CDE, pulmonary hypoplasia is often mild or absent. Even when surgical correction is required, it is typically performed electively with favorable outcomes [6]. Long-term follow-up is recommended to monitor respiratory function, diaphragmatic integrity, and lung growth [5]. It is important to note that CDE, although less severe than CDH, may coexist with other congenital anomalies, such as pulmonary sequestration, intrathoracic kidney, or non-immune hydrops, which can significantly affect the prognosis and require comprehensive perinatal planning [6–8]. Recently, Konstantinidi et al. performed a systematic review of congenital diaphragmatic eventration in the neonatal period, including 204 cases from 93 studies [3]. Their analysis mainly focused on neonatal clinical presentation, diagnostic approaches, associated anomalies, surgical management, and postnatal outcomes. Although prenatal suspicion was reported in a limited number of cases, detailed fetal imaging characteristics and the diagnostic challenges in differentiating CDE from congenital diaphragmatic hernia (CDH) were beyond the primary scope of their review. Therefore, the present literature review specifically focuses on prenatal presentation, fetal imaging findings, diagnostic pitfalls, and the role of multidisciplinary evaluation in suspected fetal CDE cases. We report a case of prenatally diagnosed congenital diaphragmatic eventration and provide a structured literature review specifically focused on prenatal imaging findings, diagnostic pitfalls, differentiation from CDH, and implications for prenatal counselling and perinatal management.

2. Materials and Methods

This case report was managed in July 2025 at the Prenatal Diagnosis Unit of Policlinico Umberto I, Rome, Italy. Clinical and radiological data were collected through a review of the maternal and neonatal medical records. Written informed consent was obtained from the patient for publication of the case and the associated clinical images. A structured literature review was conducted to identify previously reported cases of congenital diaphragmatic eventration (CDE) suspected or diagnosed during the prenatal period. This structured literature review was designed to provide a focused synthesis of prenatally suspected and diagnosed cases of congenital diaphragmatic eventration (CDE), with particular emphasis on fetal imaging findings, diagnostic pitfalls in differentiating CDE from congenital diaphragmatic hernia (CDH), and their implications for prenatal counselling and perinatal

management. Given the descriptive nature of the available evidence, the review was not intended to perform a quantitative analysis or to assess the diagnostic accuracy of imaging modalities. A literature search was performed using PubMed, Scopus, and Web of Science databases to identify relevant studies published between January 1984 and July 2025. The search strategy included the following terms: ("congenital diaphragmatic eventration" OR "diaphragmatic eventration") AND ("prenatal diagnosis" OR "fetal diagnosis" OR "antenatal diagnosis" OR "fetal imaging"). Articles were screened according to their relevance to prenatal presentation of CDE. Eligible studies were selected by two reviewers based on predefined inclusion and exclusion criteria, with any discrepancies resolved through consensus discussion. Cases were included when prenatal imaging findings were available, and the final diagnosis of CDE was confirmed by postnatal imaging, surgical findings, or autopsy. Studies reporting exclusively postnatal presentation, animal studies, and articles without sufficient prenatal information were excluded. The final literature search was performed on 31 July 2025.

3. Results

3.1. Case Presentation

A 35-year-old gravida 3, para 2 woman with a spontaneous singleton pregnancy was referred to our prenatal diagnosis unit (Policlinico Umberto I, Roma) at 20 + 2 weeks' gestation after fetal dextrocardia was suspected during routine obstetric ultrasound. Detailed fetal anatomical assessment revealed rightward displacement of the heart and an echogenic structure within the left thoracic cavity, raising suspicion of hepatic herniation. Given the concern for CDH, the lung-to-head ratio (LHR), calculated according to Jani's method [8], was greater than 50%, suggesting a favorable prognosis. No additional thoracic or abdominal abnormalities were detected. At 21 + 3 weeks' gestation, invasive prenatal testing was performed. Cytogenetic analysis demonstrated a normal female karyotype (46, XX), array-based comparative genomic hybridization (array-CGH) revealed no clinically significant copy number variations, and whole-exome sequencing (WES) did not identify pathogenic or likely pathogenic variants associated with the phenotype.

At 22 + 6 weeks' gestation, fetal magnetic resonance imaging (MRI) showed marked elevation of the left hemidiaphragm without evidence of diaphragmatic discontinuity, findings consistent with congenital diaphragmatic eventration (CDE). Cardiac dextroposition was confirmed, while cardiac anatomy and volume were normal, with a cardiothoracic ratio of approximately 1:3. Following multidisciplinary counseling, the couple chose to continue the pregnancy, with serial ultrasound examinations performed approximately every three weeks. At 30 + 5 weeks' gestation, follow-up ultrasound confirmed persistent cardiac displacement and elevation of the liver into the left thoracic cavity, with a reduction in the LHR to 22.2%, raising concern for potential pulmonary hypoplasia according to prognostic criteria used for CDH. However, the prognostic value of LHR in CDE remains uncertain and should be interpreted cautiously. Follow-up fetal MRI at 31 weeks' gestation confirmed marked elevation of the left hemidiaphragm with displacement of the left hepatic lobe, spleen, and stomach into the thorax, with compression of the left lung. Despite this, the left lung maintained a normal T2 signal, while the right lung showed preserved volume and signal intensity. Cardiac structure and volume remained normal, and no pleuropericardial effusions were detected (Figure 1).

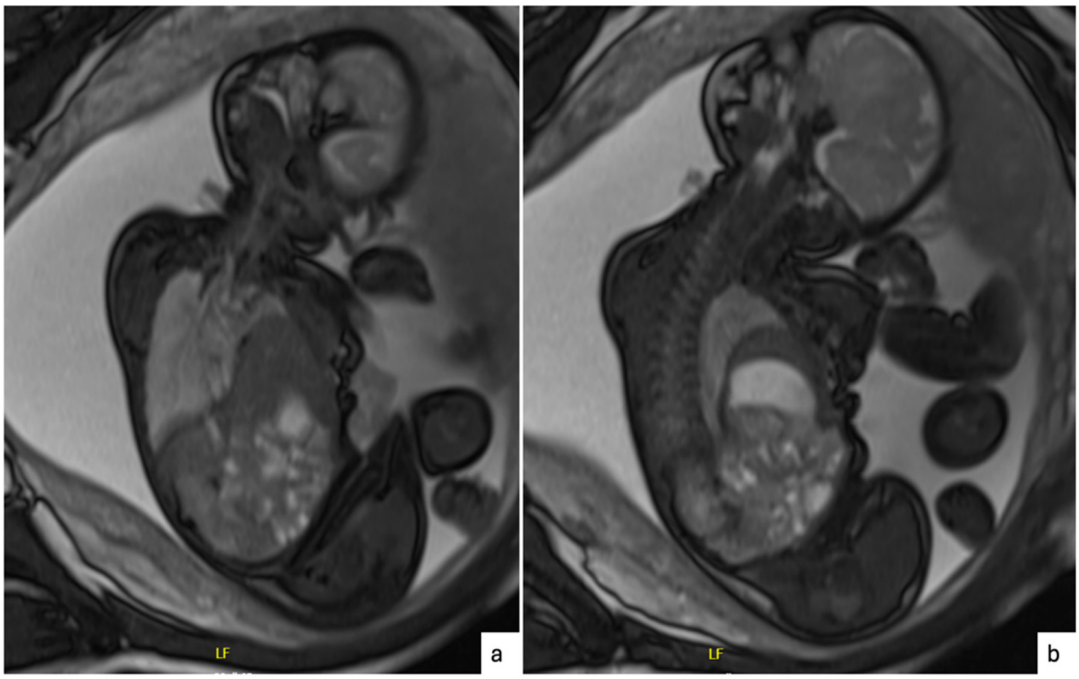


Figure 1. Fetal magnetic resonance imaging (MRI) at 31 weeks' gestation. Coronal T2-weighted image (**a**) demonstrates marked elevation of the left hemidiaphragm with preserved diaphragmatic continuity. Axial T2-weighted image (**b**) shows intrathoracic displacement of the left hepatic lobe, spleen, and stomach, associated with compression of the left lung and preserved right lung volume and signal intensity.

A multidisciplinary perinatal care plan involving obstetricians, neonatologists, pediatric surgeons, and radiologists was established. An elective cesarean delivery was planned at 38 + 6 weeks' gestation to optimize neonatal assessment and immediate access to specialized care. No additional obstetric indication for cesarean delivery was present. A female neonate was delivered, weighing 2442 g (<3rd percentile according to INTERGROWTH-21st standards), with Apgar scores of 9 and 10 at 1 and 5 min, respectively. Umbilical arterial blood gas analysis showed a pH of 7.31. At birth, the neonate presented with mild respiratory distress, initially managed with continuous positive airway pressure (CPAP) at 60% FiO_2, followed by free-flow oxygen. The newborn was subsequently transferred to the neonatal intensive care unit in spontaneous breathing, without requiring further respiratory support. On postnatal day 1, chest and abdominal radiography confirmed left-sided congenital diaphragmatic eventration (Figure 2a). Echocardiography demonstrated rightward mediastinal shift without structural cardiac anomalies, while abdominal ultrasound revealed hepatosplenomegaly (Figure 2b). Neurological examination at birth showed mild generalized hypotonia, which progressively resolved during follow-up. Subsequent neurological assessments showed normal findings, without evidence of additional abnormalities. The neonate remained clinically stable throughout hospitalization and was discharged in good condition on postnatal day 11. At six-month follow-up, the infant remained clinically stable without evidence of respiratory impairment. Continued follow-up is planned to monitor long-term respiratory development and diaphragmatic function.

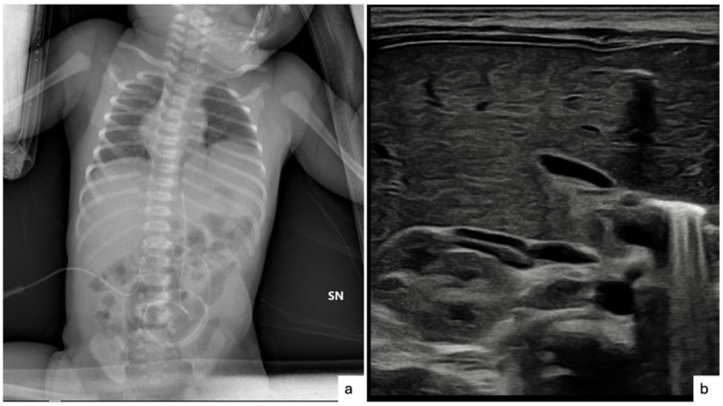


Figure 2. Postnatal imaging findings: (**a**) Chest and abdominal radiograph obtained on postnatal day 1 showing elevation of the left hemidiaphragm with rightward mediastinal shift, consistent with left-sided congenital diaphragmatic eventration; (**b**) Abdominal ultrasound demonstrating hepatosplenomegaly.

3.2. Literature Review Findings

A structured literature review was performed to identify reported cases of CDE suspected or diagnosed during the prenatal period. Seventeen relevant studies published between January 1984 and July 2025 were included, comprising a total of 19 prenatally suspected CDE cases, as some studies reported more than one case (Figure 3).

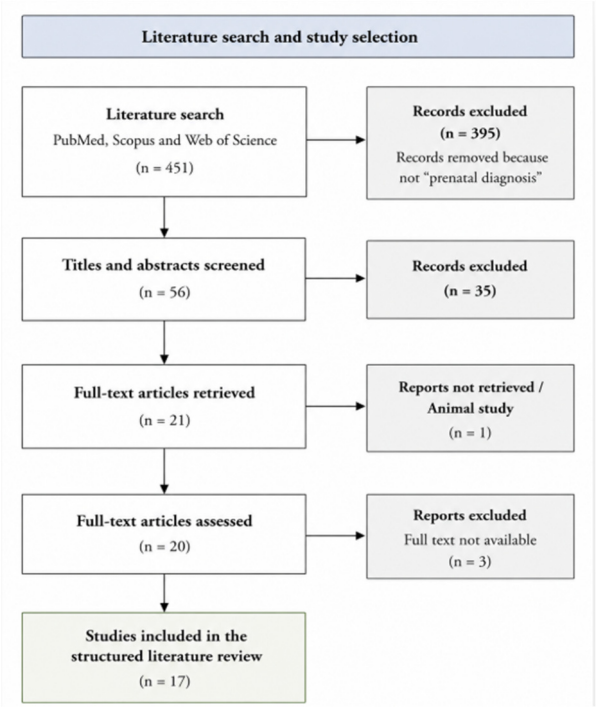


Figure 3. Flow diagram illustrating the selection process of studies included in the structured literature review of prenatally diagnosed congenital diaphragmatic eventration (CDE).

A total of 19 cases of prenatally suspected congenital diaphragmatic eventration were identified in the literature (Table 1). Prenatal imaging findings and diagnostic characteristics were analyzed in all 19 cases. However, complete postnatal outcome data were available for only 12 cases (63.2%); therefore, analyses regarding respiratory support, surgical management, and neonatal outcome were restricted to this subgroup. Among these, 17/19 cases (89.5%) were initially misdiagnosed as congenital diaphragmatic hernia. The

most frequently reported prenatal finding was intrathoracic liver (8/19, 42.1%), followed by intrathoracic stomach (5/19, 26.3%), mediastinal shift (3/19, 15.8%), and thoracic mass-like findings (2/19, 10.5%). Prenatal imaging features observed in correctly diagnosed cases included visualization of a continuous but abnormally elevated diaphragm, atypical organ displacement without evidence of a diaphragmatic defect, and associated findings such as dextrocardia or right atrial enlargement. Commonly reported misleading features included apparent intrathoracic bowel loops, polyhydramnios, and imaging findings suggestive of pulmonary hypoplasia. These findings informed subsequent analysis of imaging modalities and diagnostic performance.

Table 1. Prenatal Characteristics of Reported Cases of CDE.

Year	Author	GA at Diagnosis (Weeks)	Side	Genetic Evaluation	Imaging	Key Prenatal Findings	Misdiagnosed as CDH	Final Prenatal Diagnosis
1984 [7]	Seeds et al.	17	Bilateral	NR	US	Cardiac herniation, omphalocele	No	Correct
1986 [8]	Comstock et al.	35	Left	Trisomy 18	US	Mediastinal shift, intrathoracic stomach	Yes	Incorrect
1990 [9]	Jurcak-Zaleski et al.	29	Right	NR	US	Intrathoracic liver	Yes	Correct
1990 [10]	Thiagarajah et al.	31	Left	Normal karyotype	US	Stomach at level of heart, mediastinal shift	Yes	Correct
2001 [11]	Tsukahara et al.	35	Left	NR	US + MRI	Diaphragm continuity, intrathoracic stomach	Yes	Correct
2003 [12]	Yang JI et al.	37	Left	NR	US	Intrathoracic bowel and stomach, pulmonary hypoplasia	Yes	Incorrect
2005 [13]	Yang et al.	20	Right	Normal karyotype	US	Right atrial enlargement, intrathoracic liver, polyhydramnios	No	Incorrect
2007 [14]	Jeanty et al.	21	Right	Normal karyotype	US	Hydrops, intrathoracic liver, single umbilical artery	Yes	Incorrect
2007 [14]	Jeanty et al.	30	Central	Normal karyotype	US	Pleural and pericardial effusion, polyhydramnios	Yes	Incorrect
2007 [15]	Zankl et al.	30	Right	NR	US ± MRI	NIHF, pleural effusion, ascites, intrathoracic liver	Yes	Incorrect
2007 [15]	Zankl et al.	29–30	Right	NR	US + MRI	NIHF, polyhydramnios, intrathoracic abdominal organs	Yes	Incorrect

Table 1. *Cont.*

Year	Author	GA at Diagnosis (Weeks)	Side	Genetic Evaluation	Imaging	Key Prenatal Findings	Misdiagnosed as CDH	Final Prenatal Diagnosis
2011 [6]	Şahinoglu et al.	22	Left	Normal karyotype	US + MRI	Intrathoracic kidney, dextrocardia	Yes	Correct
2012 [16]	Cantekin et al.	17	Right	Normal karyotype	US + MRI	NIHF, pleural effusion, hepatomegaly	Yes	Correct
2019 [17]	Li Zhen et al.	12	Right	NR	US	Cystic diaphragm, intrathoracic liver	Yes	Correct
2020 [18]	De Graer et al.	22	Left	PIK3CA-related over-growth spectrum (PROS)	US + MRI	Intrathoracic double bubble sign, macrocephaly	Yes	Incorrect
2021 [19]	Sallout et al.	22–36	Left	NR	US	Multicystic thoracic mass, elevated diaphragm	Yes	Partially correct
2021 [5]	Rana et al.	24	Left	NR	US + MRI	Intrathoracic liver, mediastinal shift	Yes	Partially correct (MRI-confirmed)
2022 [20]	Mitsiakos et al.	20	Left	NR	US	Intrathoracic liver, cardiac anomalies	Yes	Incorrect
2025 [4]	Yu et al.	23	Right	NR	US	Intrathoracic liver and stomach, lung agenesis	Yes	Incorrect

Legend: NR = CDE, congenital diaphragmatic eventration; CDH, congenital diaphragmatic hernia; MRI, magnetic resonance imaging; US, ultrasound; NR, not reported. Diagnostic classification: Correct = prenatal diagnosis or suspicion of CDE. Incorrect = prenatal diagnosis of CDH or another thoracic anomaly subsequently reclassified as CDE after definitive evaluation. Partially correct = prenatal identification of a diaphragmatic abnormality without definitive differentiation between CDE and CDH until additional imaging assessment or postnatal confirmation.

Fetal magnetic resonance imaging was reported as an adjunctive diagnostic modality in seven cases. Among these cases, MRI provided additional anatomical information, particularly regarding diaphragmatic continuity and organ position, supporting diagnostic clarification in selected patients. Correct prenatal characterization was reported in 4/7 cases evaluated with MRI and in 3/12 cases evaluated with ultrasound alone. However, given the small number of cases, heterogeneity of reports, and potential publication bias, these findings should be interpreted only as an observational trend rather than evidence of superior diagnostic performance. Diagnostic inaccuracies were nevertheless reported in cases undergoing fetal magnetic resonance imaging. Genetic evaluation was reported in 9/19 cases (47.4%) across the included studies. Among these, genetic testing yielded normal results in 7/9 cases (77.8%), while abnormal findings were identified in 2/9 cases (22.2%), including trisomy 18 and PIK3CA-related overgrowth spectrum. Associated findings were variably reported across the included cases. Non-immune hydrops fetalis was described in 4/19 cases (21.1%), congenital cardiac anomalies in 2/19 cases (10.5%), and syndromic or chromosomal abnormalities in 2/19 cases (10.5%). Adverse neonatal outcomes, including early neonatal death, were reported predomi-

nantly among cases complicated by severe thoracic compression, hydrops, or additional structural abnormalities, as summarized in Table 2.

Table 2. Postnatal Management and Outcomes of Cases with Available Neonatal Follow-up Data (n = 12).

Year	Author	GA at Birth (Weeks)	Delivery	Respiratory Support	Surgery Performed	Timing of Surgery	Outcome
1986 [8]	Comstock et al.	38	Cesarean section	Mechanical ventilation	No	—	Death (2 days)
1990 [9]	Jurcak-Zaleski et al.	39	Vaginal delivery	None	No	—	Asymptomatic
1990 [10]	Thiagarajah et al.	39	Vaginal delivery	Mild tachypnea	No	—	Asymptomatic
2001 [11]	Tsukahara et al.	39	Vaginal delivery	Supplemental oxygen (24 h)	No	—	Asymptomatic
2003 [12]	Yang JI et al.	38	Vaginal delivery	Mechanical ventilation	Yes	24 h	Good outcome
2005 [13]	Yang et al.	41	Cesarean section	Mechanical ventilation	Yes	7 days	Good outcome
2007 [15]	Zankl et al.	30	Cesarean section	Mechanical ventilation (ineffective)	No	—	Death (1 h)
2007 [15]	Zankl et al.	30	Cesarean section	No resuscitation	No	—	Death (1 h)
2012 [16]	Cantekin et al.	37	Cesarean section	CPAP followed by supplemental oxygen	No	—	Asymptomatic
2020 [19]	Sallout et al.	37 + 3	Vaginal delivery	Mechanical ventilation → nasal cannula	Yes	6 months	Good outcome
2021 [5]	Rana et al.	37+	Vaginal delivery	Supplemental oxygen	Yes	16 months	Good outcome
2022 [20]	Mitsiakos et al.	32	Cesarean section	Mechanical ventilation	Yes	Neonatal	Death (40 h)

Notes: "→" indicates a switch from mechanical ventilation to nasal cannula.

Among the 12/19 cases (63.2%) with available postnatal outcome data, respiratory support was required in 9/12 neonates (75.0%). Minimal respiratory support, including supplemental oxygen or short-term continuous positive airway pressure, was reported in 3/12 cases (25.0%), whereas invasive mechanical ventilation was required in 6/12 cases (50.0%). Early neonatal death secondary to severe respiratory failure occurred in 3/12 cases (25.0%), all involving preterm neonates. Surgical intervention was performed in 5/12 cases (41.7%), whereas conservative management was reported in 7/12 cases (58.3%). Among surgically treated neonates, favorable outcomes were observed in 4/5 cases (80.0%), while adverse outcomes were reported in 1/5 cases (20.0%). Among conservatively managed cases, favorable outcomes were described in 4/7 neonates (57.1%), whereas adverse outcomes occurred in 3/7 cases (42.9%). Delayed surgical repair beyond the neonatal period was reported in 2/5 surgically managed cases (40.0%), performed at 6 and 16 months of age, respectively.

4. Discussion

Congenital diaphragmatic eventration is a rare anomaly involving abnormal elevation of the diaphragm due to partial or complete replacement of muscular tissue by fibroelastic connective tissue. Although asymptomatic postnatally, when diagnosed in utero, it can mimic congenital diaphragmatic hernia (CDH), which has significantly different manage-

ment and prognosis. Accurate differentiation among congenital diaphragmatic anomalies is essential, as prognosis and management differ substantially. While CDH involves a true diaphragmatic defect with intrathoracic herniation and is often associated with poor outcomes, CDE is characterized by an intact but elevated diaphragm and generally carries a more favorable and variable prognosis. These distinctions highlight the critical role of prenatal imaging in assessing diaphragmatic continuity and organ position to ensure accurate diagnosis, as shown in Table 3.

Table 3. Prenatal imaging features useful for differential diagnosis between congenital diaphragmatic eventration (CDE) and congenital diaphragmatic hernia (CDH).

Diagnosis	Diaphragm	Herniation	Liver Position	Key Prenatal Imaging Feature	Prognosis
CDH	Defective	Present	Intrathoracic	Diaphragmatic discontinuity with abdominal organ herniation	Often poor
CDE	Intact	Absent	Elevated	Continuous diaphragm with abnormal elevation	Variable, often good
Diaphragm agenesis	Absent	Present	Intrathoracic	Complete absence of diaphragm elevation	Very poor

The role of fetal MRI in the prenatal assessment of CDE has been previously described. Tsukahara et al. [11] first demonstrated the usefulness of fetal MRI in identifying diaphragmatic continuity and supporting differentiation between CDE and CDH, while Rana et al. [5] reported the value of MRI in clarifying the diagnosis in a case initially suspected as CDH based on ultrasound findings. Consistent with these previous observations, fetal MRI may represent a useful complementary imaging modality in selected cases with uncertain ultrasound findings by providing additional anatomical information regarding diaphragmatic continuity, organ position, and the relationship between abdominal and thoracic structures. In the present literature review, MRI contributed to correct or partially correct prenatal characterization in four reported cases (Tsukahara et al. [11], Şahinoglu et al. [6], Cantekin et al. [16], and Rana et al. [5]), particularly when ultrasound findings were inconclusive or misleading. However, MRI did not completely eliminate diagnostic uncertainty. Considering the small number of cases, heterogeneity of reports, differences in imaging protocols, and potential publication bias, these observations should be interpreted as a descriptive trend rather than evidence of superior diagnostic performance compared with ultrasound alone.

An additional relevant aspect emerging from the literature concerns the genetic background of CDE. In most reported cases, genetic testing was either not performed or revealed normal results, supporting the hypothesis that CDE is frequently an isolated developmental anomaly. However, in a minority of cases, CDE has been associated with chromosomal abnormalities or syndromic conditions, including trisomy 18 and PIK3CA-related overgrowth spectrum (PROS). These findings suggest that, although uncommon, CDE may occur as part of a broader phenotypic spectrum. In our case, comprehensive genetic evaluation, including karyotype, array-CGH, and whole-exome sequencing, did not identify any pathogenic variants, further supporting an isolated form of CDE. Therefore, detailed genetic assessment should be considered, particularly in the presence of additional anomalies, to refine diagnosis, guide counseling, and exclude syndromic associations.

Another important observation is the heterogeneity of associated findings and the final prognosis. While some cases presented as isolated anomalies, others were associated with complex conditions such as non-immune hydrops fetalis (NIHF), congenital heart anomalies, or genetic syndromes. Cases with NIHF or severe thoracic compression ap-

peared to have a worse prognosis, suggesting that the clinical spectrum of CDE ranges from mild, asymptomatic forms to severe presentations associated with pulmonary hypoplasia and poor neonatal outcomes, as also shown in Table 2. In the present case, several elements initially suggested a diagnosis of CDH, including cardiac dextroposition and apparent intrathoracic liver. However, the relatively preserved early LHR >50% and the absence of additional structural anomalies were atypical for severe CDH. The progressive reduction in LHR during follow-up raised concern for pulmonary hypoplasia, but fetal MRI consistently demonstrated diaphragmatic continuity, ultimately supporting the diagnosis of CDE. Importantly, despite significant thoracic organ displacement and compression of the left lung, the preservation of normal T2 signal intensity suggested maintained lung parenchymal integrity. This radiological-functional dissociation is particularly relevant, as it may explain the relatively mild postnatal course observed in our case.

Unlike CDH, where pulmonary hypoplasia is often severe and clinically limiting, our patient required only transient respiratory support and did not need surgical intervention, showing a favorable short-term clinical course at the six-month follow-up. These findings are consistent with previously reported cases in which preserved lung signals on MRI correlated with favorable outcomes, even in the presence of marked anatomical distortion. Therefore, our case supports the importance of assessing diaphragmatic integrity and lung characteristics, in addition to organ position, as part of prenatal evaluation and counselling. However, longer follow-up is required to better define the prognostic significance of these findings. The integration of serial ultrasound and fetal MRI, within a multidisciplinary framework, allows a more precise distinction between CDE and CDH, ultimately improving parental counseling and perinatal management.

The discrepancy observed in our case also highlights a broader issue emerging from the available literature. Although prenatal diagnosis of CDE has become increasingly feasible, our review demonstrates that disease-specific evidence guiding clinical management rapidly declines once the diagnosis has been established. Unlike congenital diaphragmatic hernia (CDH), for which structured multidisciplinary frameworks are available for prenatal counselling, antenatal surveillance, prognostic assessment, neonatal management, and long-term follow-up [21–23], no equivalent disease-specific recommendations currently exist for CDE. Consequently, counselling and clinical management remain largely individualized and are frequently extrapolated from CDH despite the important differences in pathophysiology and prognosis between the two conditions. Our case illustrates this challenge, as a markedly reduced LHR was associated with only mild neonatal respiratory compromise, emphasizing the uncertainty surrounding CDH-derived prognostic indices in CDE and the need for disease-specific prognostic and management studies.

Overall, neonatal presentation of CDE appears highly variable, ranging from asymptomatic cases to severe respiratory compromise requiring intensive care. Reported outcomes suggest that prognosis is mainly influenced by the severity of pulmonary compromise, prematurity, and the presence of associated anomalies rather than by diaphragmatic eventration alone. Surgical intervention should therefore be individualized according to clinical presentation, as some patients remain stable with conservative management whereas others may require diaphragmatic plication because of persistent respiratory symptoms. Importantly, the need for surgery itself does not necessarily indicate an unfavorable prognosis, as favorable outcomes have been described after appropriate surgical correction.

Despite this variability, surgical intervention was not required in more than half of reported cases with available postnatal follow-up data (7/12, 58.3%), particularly in those who remained asymptomatic or only mildly symptomatic. Conversely, surgery was performed in 5/12 cases (41.7%), generally in the presence of persistent respiratory symptoms or clinical deterioration. Among surgically managed patients, outcomes were favorable in

most cases (4/5, 80%), and good results were also observed when diaphragmatic plication was delayed for several months after birth, as shown in two cases [4,12] operated at 6 and 16 months, respectively. An important observation is that the need for surgery does not necessarily correlate with poor prognosis. In fact, neonates undergoing surgical correction often achieved favorable outcomes (4/5, 80%), suggesting that intervention can be effective when appropriately indicated. In contrast, poor outcomes were observed in both surgically and non-surgically managed patients (3/7, 42.9% vs. 1/5, 20%) and were predominantly associated with severe clinical presentations rather than with the diaphragmatic abnormality itself. These findings further emphasize the importance of distinguishing CDE from CDH in the prenatal setting. Misdiagnosis may lead to inaccurate prognostic assessment, as CDH is typically associated with higher mortality and more aggressive perinatal management. In contrast, CDE—particularly in isolated forms—often follows a more favorable clinical course and may not require immediate surgical intervention.

Taken together, these findings suggest that prognosis in CDE is primarily determined by the extent of pulmonary compromise and associated anomalies, rather than by the diaphragmatic abnormality itself. Accordingly, a comprehensive approach integrating prenatal imaging and postnatal clinical assessment is essential to guide management and optimize outcomes.

This study has several limitations. First, the number of reported prenatal CDE cases is limited due to the rarity of the condition, and the available evidence is derived mainly from individual case reports. Second, publication bias cannot be excluded, as unusual or more complex cases may be more likely to be reported. Third, the retrospective nature of the literature review and the heterogeneity among published cases, including differences in prenatal imaging protocols, diagnostic approaches, genetic evaluation, and postnatal follow-up, limit direct comparisons between cases. Furthermore, the comparison between ultrasound and fetal MRI should be interpreted only as a descriptive observation, as the available data do not allow assessment of diagnostic accuracy or superiority of one imaging modality over another. Finally, the follow-up duration of the presented case remains limited; therefore, longer clinical surveillance is necessary to evaluate respiratory development and long-term outcomes. Larger multicenter studies with standardized reporting are required to better define prenatal diagnostic criteria and prognostic factors.

5. Conclusions

Congenital diaphragmatic eventration remains an exceptionally rare prenatal condition for which evidence guiding clinical management is still lacking. Although prenatal differentiation from congenital diaphragmatic hernia is increasingly achievable, disease-specific recommendations for counselling, prognostic assessment, antenatal surveillance, obstetric management, neonatal care, and long-term follow-up are currently unavailable. By combining a new prenatally diagnosed case with a structured review of the prenatal literature, our study highlights that the current challenge in CDE is no longer establishing the diagnosis itself, but defining evidence-based management once the diagnosis has been made. Multicentre collaborative studies will be essential to develop disease-specific recommendations for counselling and clinical management.

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Informed Consent Statement: Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

Data Availability Statement: The data supporting the findings of this study are available from the corresponding author upon reasonable request.

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Conflicts of Interest: The authors declare no conflicts of interest.

Abbreviations

The following abbreviations are used in this manuscript:

CDE	Congenital diaphragmatic eventration
CDH	Congenital diaphragmatic hernia
MRI	Magnetic resonance imaging
LHR	Lung-to-head ratio
NIHF	Non-immune hydrops fetalis
CPAP	Continuous positive airway pressure
WES	Whole-exome sequencing
Array-CGH	Array comparative genomic hybridization
PROS	PIK3CA-related overgrowth spectrum

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