

A Diagnostic Dilemma: Congenital Diaphragmatic Hernia versus Diaphragmatic Eventration in a Neonate—A Case Report

Ezioma Anne Alinnor*, Idorenyin Diamond, Chioma Ada Nnah, Kelechi Cynthia Nwokoro

Department of Paediatrics, University of Port Harcourt Teaching Hospital, Port Harcourt, Rivers State, Nigeria

Email: *alinnorezioma@gmail.com

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Abstract

Background: Congenital diaphragmatic anomalies encompass a spectrum of developmental defects, with congenital diaphragmatic hernia (CDH) and congenital diaphragmatic eventration (CDE) representing the most clinically significant variants. These diaphragmatic abnormalities may present with neonatal respiratory distress, intrathoracic abdominal viscera, and mediastinal shift. While CDH involves a true diaphragmatic defect with herniation of abdominal organs into the thorax, CDE is characterized by an intact but thinned, elevated diaphragmatic sheet. Preoperative differentiation remains challenging, particularly in settings lacking advanced imaging modalities. **Case Presentation:** We report a male infant referred with progressive respiratory distress and cyanosis from birth. Initial chest radiography revealed gas-filled bowel loops in the left hemi-thorax with mediastinal shift, prompting a presumptive CDH diagnosis. Computed tomography demonstrated elevation of the left hemidiaphragm with intrathoracic stomach and bowel loops and right-ward mediastinal shift. Although diaphragmatic continuity was suggested on imaging, the distinction between CDH and CDE remained uncertain preoperatively. Exploration thoracotomy at 8 weeks demonstrated an intact, diffusely elevated, thinned left hemidiaphragm without a true diaphragmatic defect, confirming CDE. Diaphragmatic plication was performed, with satisfactory clinical improvement. **Conclusion:** This case reveals the overlap between CDH and CDE and the importance of multimodal imaging, including dynamic ultrasonography when available, to enhance diagnostic accuracy. In settings where advanced imaging is unavailable, or inconclusive, operative findings may establish the diagnosis when surgery is clinically indicated.

Keywords

Congenital Diaphragmatic Eventration, Congenital Diaphragmatic Hernia, Neonatal Respiratory Distress, Diaphragmatic Plication, Diagnostic Imaging, Low-Resource Settings

1. Background

Congenital diaphragmatic anomalies represent a heterogeneous group of developmental defects with profound implications for neonatal respiratory function and long-term outcomes. Congenital diaphragmatic hernia (CDH), occurring in approximately 1 in 2000 - 3600 live births, results from incomplete fusion of the pleuroperitoneal folds during embryogenesis, permitting abdominal contents to herniate into the thoracic cavity [1]. It is associated with pulmonary hypoplasia, pulmonary vascular remodeling, pulmonary hypertension, and cardiac dysfunction, which contribute substantially to neonatal morbidity and mortality [1].

Congenital diaphragmatic eventration (CDE) is a distinct condition characterized by an anatomically continuous but abnormally elevated diaphragm due to incomplete muscular development and replacement with fibroelastic tissue. It may cause respiratory compromise through reduced lung expansion and mediastinal displacement [2].

The diagnostic challenge arises from overlapping clinical presentations, as both conditions may manifest with tachypnea, cyanosis, decreased breath sounds, and abdominal auscultation in the hemithorax [2]. Distinguishing CDE from CDH before surgery is clinically important because the operative findings, surgical approach, prognosis, and counselling differ [2]. Imaging assessment should focus on diaphragmatic continuity, contour, position, and motion. However, overlap in imaging appearances means that a definitive distinction may not always be possible before surgery [2].

We report a neonate whose clinical and radiological findings were initially interpreted as CDH but whose operative findings confirmed congenital diaphragmatic eventration. The case illustrates the diagnostic challenge posed by congenital diaphragmatic abnormalities, particularly where advanced imaging modalities are not readily available.

2. Case Presentation

2.1. Patient Information and Antenatal History

A male infant, born on March 16, 2026, was referred from an out-of-state facility to our tertiary neonatal center on day 5 of life, but presented on day 7 of life. He was the firstborn of a monogamous family with no family history of congenital anomalies or similar conditions in first-degree relatives. Antenatal care commenced at 7 weeks' gestation, with three routine prenatal ultrasound scans performed reported no fetal abnormalities. The pregnancy was uncomplicated, with no mater-

nal illness, diabetes, or teratogenic risk exposure.

2.2. Birth and Initial Course

Labor onset was spontaneous, and he was delivered vaginally at term. Birth weight was 3.5 kg (appropriate for gestational age). Immediately after birth, he developed progressive breathlessness and intermittent cyanosis, that persisted despite supplemental oxygen administration. Initial management at the referring facility was for presumed respiratory distress syndrome with broad-spectrum antibiotics and oxygen therapy, without clinical improvement.

2.3. Initial Diagnostic Findings at the Referral Facility

Chest radiography performed at the referring facility demonstrated gas-filled bowel loops occupying the left hemithorax with significant mediastinal shift to the right (**Figure 1**). Electrocardiography showed right axis deviation and an apparent rightward cardiac position, attributed to mediastinal displacement rather than primary dextrocardia. Persistent respiratory deterioration and the absence of definitive paediatric surgical facilities prompted transfer to our tertiary center.

2.4. Clinical Examination at the Tertiary Center

On admission at 7 days of life, the infant had severe respiratory distress with a respiratory rate of 90 breaths/minute; heart rate: 168 beats/minute; and central cyanosis despite oxygen supplementation. Other findings included a displaced apex beat at the right fifth intercostal space, audible bowel sounds on auscultation of the left hemithorax and reduced breath sounds on the left side.

2.5. Advanced Imaging and Laboratory Studies

Chest Computed Tomography demonstrated marked elevation of the left hemidiaphragm with the stomach and bowel loops within the left hemithorax, and associated rightward mediastinal shift (**Figure 2**). These findings raised a preoperative suspicion in favour of a CDH, however, the imaging findings did not conclusively distinguish a diaphragmatic defect from congenital diaphragmatic eventration.

Echocardiography: 2-D echocardiography revealed a patent ductus arteriosus (PDA) with left-to-right shunt; Type III right-to-left interatrial septal aneurysm; Mild tricuspid regurgitation and Mild left ventricular hypertrophy.

2.6. Laboratory Investigations

Complete blood count: Hemoglobin 14 g/dL, RBC $4.4 \times 10^{12}/L$, WBC $8.8 \times 10^9/L$ (neutrophils 46%, lymphocytes 52%, eosinophils 2%), Platelets $370 \times 10^9/L$.

Renal function: Serum urea 2.3 mmol/L, Creatinine 35 $\mu\text{mol}/L$, Sodium 137 mmol/L, Potassium 4.4 mmol/L, Bicarbonate 20 mmol/L.

Serology: Hepatitis B surface antigen, hepatitis C virus, and rapid plasma reagin were all seronegative.

Dynamic ultrasonography and magnetic resonance imaging (MRI) were not

performed.

2.7. Surgical Intervention and Outcome

Surgical repair was delayed for 7 weeks due to constraints of logistics. At 8 weeks of life, the infant underwent exploratory thoracotomy. Intraoperative findings revealed:

No true diaphragmatic defect; the diaphragm was anatomically intact.

Diffuse elevation of the left hemidiaphragm with marked thinning.

Muscular laxity affecting the left dome and central aspects.

No associated thoracoabdominal anomalies or intrathoracic adhesions.

The diagnosis of congenital diaphragmatic eventration was confirmed intraoperatively. Diaphragmatic plication was performed with excision of redundant diaphragmatic tissue to achieve taut repositioning. The procedure was completed without complications.

2.8. Postoperative Course and Follow-Up

The infant was managed in the intensive care unit for 5 days postoperatively.

Extubation: Within 24 hours of surgery, with oxygen saturation 98% on supplemental oxygen.

Respiratory status: No respiratory compromise; oxygen requirement resolved by postoperative day 2.

Nutrition: Oral breastmilk feeding commenced on postoperative day 3.

Chest tube: Removed on post-operative day 5.

Ward transfer: Postoperative day 5.

Discharge: Postoperative day 7 in stable condition.

Postoperative Imaging: Chest radiography following extubation demonstrated well-defined left diaphragmatic contours, re-expansion of the left lung field with incomplete apical expansion, and resolution of mediastinal shift (**Figure 3**).

Follow-up: At 20 days' post-discharge, the infant remained stable with respiratory rate 50 breaths/minute, no respiratory symptoms, and adequate weight gain. Physical examination revealed no residual abnormalities.



Figure 1. Pre-operative chest radiograph demonstrating gas-filled bowel loops occupying the left hemithorax with rightward mediastinal shift, initially suggestive of congenital diaphragmatic hernia.

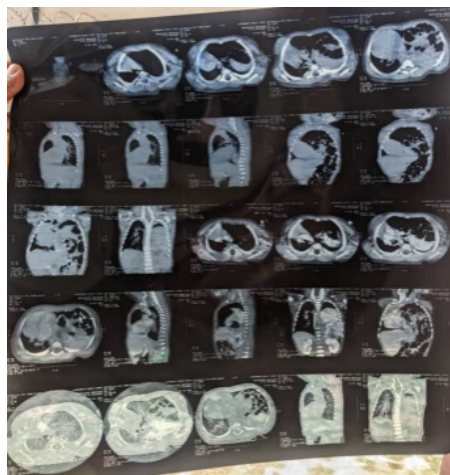


Figure 2. Pre-operative axial CT scan showing marked elevation of the left hemidiaphragm with intrathoracic stomach and bowel loops, mediastinal shift to the right, and preserved diaphragmatic continuity suggestive of eventration.

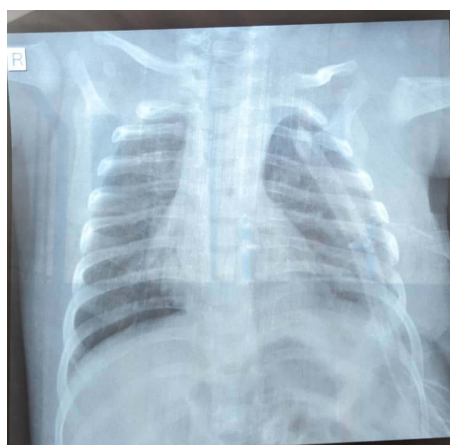


Figure 3. Chest radiography following extubation demonstrated well-defined left diaphragmatic contours, re-expansion of the left lung field with incomplete apical expansion, and resolution of mediastinal shift.

3. Discussion

3.1. Embryology, Pathophysiology and Imaging Differentiation: CDH versus CDE

Congenital diaphragmatic hernia (CDH) results from abnormal diaphragm development, creating a defect through which abdominal viscera may herniate into the thoracic cavity. The reduced intrathoracic space may impair lung development and is associated with pulmonary hypoplasia, pulmonary vascular abnormalities, pulmonary hypertension, and cardiac dysfunction [1] [3]. Diaphragm development is complex, and disruption of normal developmental processes contributes to the spectrum of congenital diaphragmatic defects [2].

Congenital diaphragmatic eventration (CDE) differs from CDH because the diaphragm is anatomically continuous. In CDE, deficient muscular development or marked thinning of the diaphragm causes abnormal elevation of the hemidiaphragm. This may reduce ipsilateral lung expansion and displace mediastinal structures, producing respiratory symptoms that can resemble CDH [4] [5].

The distinction is clinically important because CDH involves a true diaphragmatic defect, whereas CDE is managed by plication of an intact but lax diaphragm when symptoms are clinically significant. In the present case, the initial clinical and radiological findings suggested CDH; however, operative exploration demonstrated an intact, diffusely elevated left hemidiaphragm, confirming CDE.

Chest radiography may show diaphragmatic elevation, intrathoracic abdominal viscera, lung compression, and mediastinal shift in both conditions. Consequently, radiography alone may not reliably distinguish CDE from CDH [6]. Cross-sectional imaging can assist by evaluating diaphragmatic continuity, contour, and the relationship of abdominal organs to the diaphragm. In CDE, imaging may show a smooth, elevated but continuous diaphragm, whereas CDH may demonstrate a focal defect with herniation of abdominal contents. Nevertheless, overlap in imaging appearances may occur, especially when the diaphragm is markedly attenuated or poorly visualised [6].

Computed tomography in this case demonstrated marked elevation of the left hemidiaphragm, intrathoracic stomach and bowel loops, and rightward mediastinal shift. Although diaphragmatic continuity was suggested, the findings did not conclusively distinguish CDE from CDH before surgery. Dynamic ultrasonography can provide useful functional assessment by evaluating diaphragmatic excursion and may support the diagnosis of diaphragmatic dysfunction when reduced or paradoxical movement is demonstrated. Its findings should be interpreted with anatomical imaging and the clinical context, as no single modality is definitive in every case [7].

3.2. Prenatal Diagnosis and Clinical Presentation

Prenatal ultrasonography can identify CDH by demonstrating intrathoracic abdominal organs, an abnormal cardiac position, and mediastinal displacement. Prenatal recognition supports counselling, assessment for associated anomalies, and planning for delivery in a centre with appropriate neonatal and paediatric surgical support [8]. Prenatal recognition of CDE may be more difficult because the diaphragm remains intact and the imaging appearance can overlap with CDH. In this case, routine antenatal ultrasonography reportedly did not identify a fetal abnormality; therefore, a normal antenatal ultrasound examination does not necessarily exclude congenital diaphragmatic abnormalities.

Both CDH and CDE may present in the neonatal period with respiratory distress, cyanosis, reduced breath sounds on the affected side, bowel sounds within the thorax, and mediastinal displacement. In CDH, severity is strongly influenced by pulmonary hypoplasia, pulmonary vascular disease, and associated anomalies;

in CDE, respiratory compromise is more directly related to diaphragmatic elevation and reduced lung expansion [1] [4] [5]. In this infant, the presence of bowel sounds in the left hemithorax, reduced left-sided breath sounds, and rightward displacement of the cardiac apex raised concern for a congenital diaphragmatic abnormality but did not distinguish CDH from CDE.

3.3. Associated Anomalies

CDH may occur as an isolated defect or with structural, chromosomal, or genetic abnormalities. Careful physical examination, echocardiographic assessment, and consideration of genetic evaluation are appropriate when additional anomalies are identified [1] [9]. In contrast, CDE may occur as an isolated abnormality or alongside other conditions; evaluation should therefore be guided by the infant's clinical findings rather than assuming that CDE shares the same anomaly profile as CDH [4].

In this case, echocardiography identified a patent ductus arteriosus, type III interatrial septal aneurysm with right-to-left shunting, mild tricuspid regurgitation, and mild left ventricular hypertrophy. These findings were considered during respiratory and peri-operative assessment.

3.4. Surgical Management and Pulmonary Implications

Management of CDH requires initial physiological stabilization, careful respiratory support, and assessment of pulmonary hypertension and cardiac function before repair [3] [10]. For symptomatic CDE, diaphragmatic plication is an established treatment intended to flatten and stabilise the elevated diaphragm, reduce lung compression, and improve respiratory mechanics [5]. The choice of open or minimally invasive plication should be individualised according to patient stability, anatomy, available expertise, and local resources [11].

In the present case, persistent respiratory symptoms and unresolved preoperative diagnostic uncertainty supported surgical exploration after clinical stabilisation. Intraoperatively, the left hemidiaphragm was intact but diffusely elevated and lax, without a true diaphragmatic defect, confirming CDE. Diaphragmatic plication was therefore performed.

The respiratory consequences of CDH are primarily related to impaired lung development, pulmonary vascular abnormalities, pulmonary hypertension, and cardiac dysfunction [1] [3]. In CDE, respiratory compromise is more directly related to mechanical elevation of an intact diaphragm, reduced ipsilateral lung expansion, and mediastinal displacement [4] [5]. Although both conditions may cause neonatal respiratory distress, their different anatomical substrates have important implications for operative planning and counselling.

3.5. Diagnostic Challenges in Low Resource Settings

In settings where advanced imaging, dynamic ultrasonography, paediatric intensive care, and specialised paediatric surgical services are limited, early recognition

of respiratory distress and prompt referral to an appropriate centre remain important. Basic chest radiography may identify intrathoracic bowel loops, diaphragmatic elevation, or mediastinal shift but may not reliably distinguish CDH from CDE [6].

When imaging remains inconclusive, operative exploration may establish the diagnosis when surgery is clinically indicated. This should not be interpreted as a requirement for exploration solely for diagnostic purposes; the decision should depend on the infant's clinical condition, available diagnostic resources, and the anticipated benefit of surgical treatment.

4. Limitations

A limitation of this case was the inability to establish a definitive distinction between CDH and CDE before surgery. Although computed tomography suggested diaphragmatic continuity, the imaging findings remained equivocal. Dynamic ultrasonography and magnetic resonance imaging were not available. The diagnosis of CDE was ultimately confirmed intraoperatively.

As a single case report, this report cannot establish the diagnostic accuracy of any imaging modality or determine comparative outcomes of different surgical approaches. Follow-up duration should also be stated accurately and interpreted cautiously.

Despite these limitations, this case provides valuable insights into the diagnostic challenge of CDE in settings lacking advanced imaging modalities.

4.1. Ethical Considerations

Informed consent was obtained from the parents for publication of this case report and accompanying images. In obtaining consent, it was explicitly stated that all identifying information would be removed to maintain patient confidentiality, and parents were assured that participation would not affect the child's medical care.

4.2. Clinical Relevance and Learning Points

This case has important implications for clinical practice, particularly in low-resource settings:

- CDH and CDE may present with similar neonatal respiratory and radiographic findings, including intrathoracic abdominal viscera and mediastinal shift.
- Assessment of diaphragmatic continuity, contour, and motion, where available, can assist in distinguishing CDE from CDH; however, imaging may remain inconclusive in some cases.
- CDE is characterised by an intact but elevated diaphragm, whereas CDH involves a true diaphragmatic defect.
- When symptoms are significant, diaphragmatic plication is an established treatment for CDE.

- In settings with limited diagnostic resources, referral, multidisciplinary assessment, and surgery when clinically indicated may be necessary to establish the diagnosis and provide treatment.

5. Conclusions

Congenital diaphragmatic eventration represents a rare but clinically significant mimic of congenital diaphragmatic hernia, with overlapping clinical and radiographic features that pose substantial diagnostic challenges, particularly in settings lacking advanced prenatal imaging and diagnostic modalities. Congenital diaphragmatic eventration should be considered in neonates with respiratory distress, intrathoracic abdominal viscera, and mediastinal shift, particularly when imaging suggests diaphragmatic continuity. Although CT and ultrasonography can provide useful anatomical and functional information, they may not conclusively distinguish CDE from CDH in every case. In this patient, surgical exploration established the diagnosis of CDE and enabled diaphragmatic plication.

Careful interpretation of clinical and imaging findings, together with timely referral and multidisciplinary management, is important for optimising care in both well-resourced and resource-limited settings.

Author's Contribution

Alinnor AE: Conception and design, data acquisition, surgical management, manuscript drafting, critical revision.

Diamond I: Data acquisition, clinical management, manuscript revision.

Nnah CA: Data acquisition, postoperative care, manuscript revision.

Nwokoro KC: Data acquisition, follow-up care, manuscript revision.

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Conflicts of Interest

The authors declare no conflicts of interest, financial or otherwise, related to the publication of this case report.

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