

Late-diagnosed left congenital diaphragmatic hernia at term with extensive bowel herniation and favorable outcome: A case report

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Background

Congenital diaphragmatic hernia (CDH) is a developmental defect of the diaphragm that allows abdominal viscera to herniate into the thoracic cavity. More than half of CDH cases are suspected prenatally on routine anomaly scanning between 18 and 24 weeks of gestation. However, missed or delayed diagnosis can occur and may significantly complicate perinatal management.

Prenatal Course

Maternal background

33-year-old (G4P1A2L1, IVF). PCOS. No underlying disease, or infectious complications.

20 weeks	Routine anomaly scan — no abnormalities detected
34 weeks	Retrospective review: suspicious thoracic lesion first noted
39+2 wks	Dextroposition of fetal heart; herniated bowel loops in left chest → referred to tertiary center

Prenatal Ultrasound Findings at 39+5 weeks

- Lung-to head ratio (LHR) 2.4 (O/E LHR 51.4%); left CDH with herniated bowel 8.0×4.2 cm
- Dextroposition + cardiac axis 16°; RV 1.5 cm >> LV 0.8 cm
- Hypoplastic aortic isthmus (0.36 cm) — squeezed aorta

Prenatal Ultrasound Images (GA 39+5 weeks)



Figure 1
Abdominal view: stomach (st) and spleen (sp) remain intra-abdominal; bowel loops (bowel) and left lung (lt) visible in the thorax

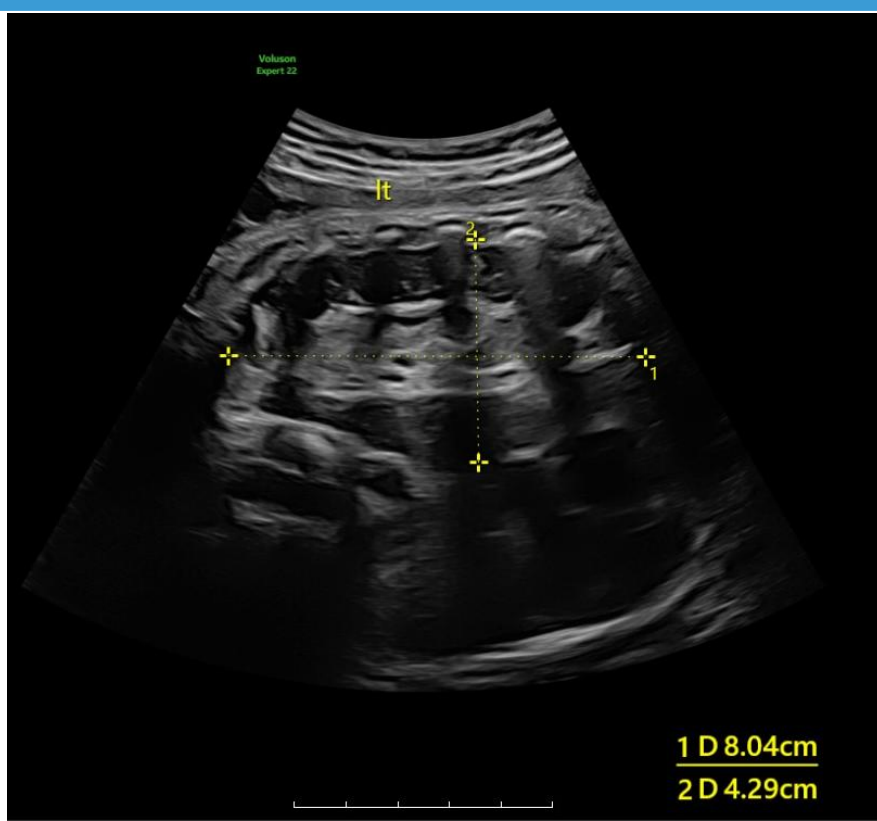


Figure 2
Measurement of herniated bowel mass in left thorax: 8.04 × 4.29 cm

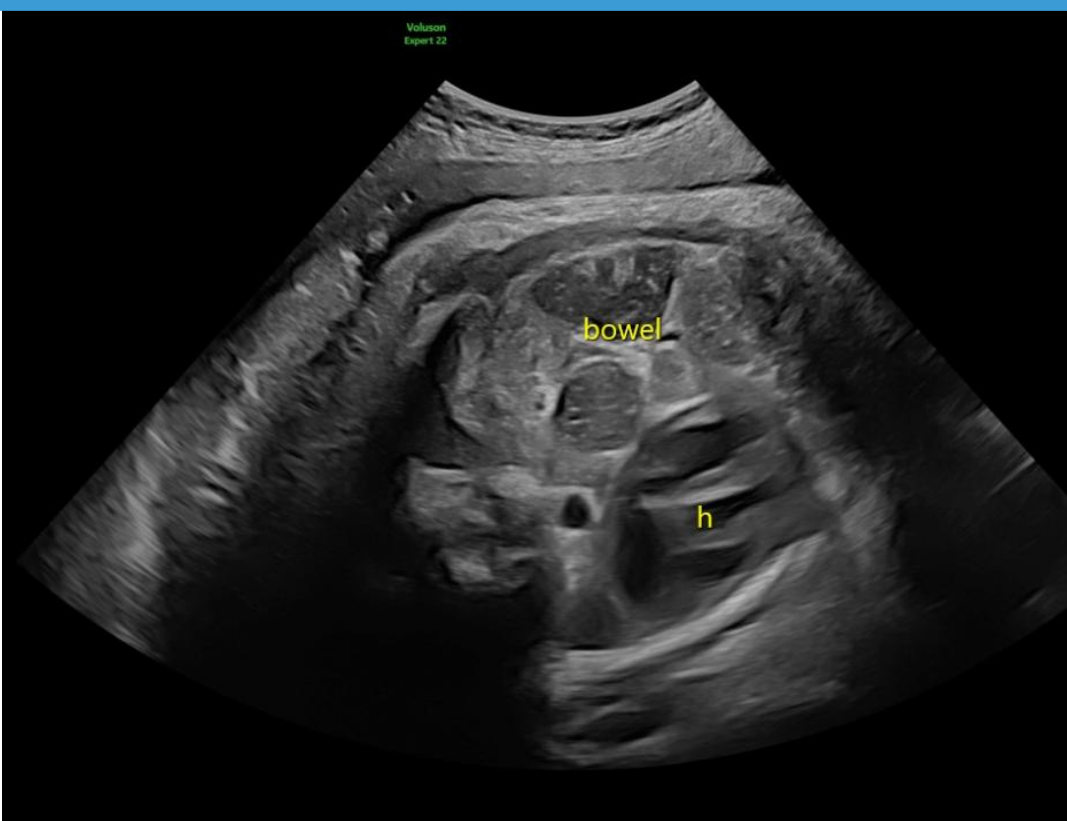


Figure 3
Four-chamber view showing dextroposition of the heart (h) with bowel loops occupying the left thoracic cavity

Delivery

Cesarean delivery at 39+6 weeks (Jan 13, 2026). EXIT procedure performed — intubation prior to cord clamping under multidisciplinary team.

Neonate: male, 3,880 g (80th %), Apgar 5/7. No meconium staining, no gross anomaly.

Maternal patient was discharged uneventfully on postoperative day 4.

Neonatal Diagnosis

- Left-sided congenital diaphragmatic hernia (CDH)
- Persistent pulmonary hypertension of the newborn (PPHN) — severe
- Patent ductus arteriosus (PDA) — huge, 6–9 mm, R→L shunt
- Patent foramen ovale (PFO) / ASD secundum
- Hypoplastic aorta with squeezed isthmus
- R/O Hypoxic-ischemic encephalopathy (HIE)

Surgical Repair — On postnatal day 2 (Jan 14)

Defect	2.5 × 1.5 cm
Herniated	Entire small bowel + near-total large bowel
In abdomen	Stomach & spleen in situ
Closure	Ethibond #2-0 interrupted; lateral stitch anchored to rib
EBL	3 cc — no drain, no complications

NICU Clinical Course

HD 1	EXIT intubation → NICU. HFOV, iNO, inotropics (Dopamine, HCS). PPHN moderate–severe
HD 2	CDH primary repair. Post-op RL shunt worsened → max sedation, max inotropics
HD 3	Suprasystemic PPHN (PaO ₂ 43). Remodulin started (self-pay) as ECMO-salvage strategy
HD 7	Mode changed to ACMV. PDA closed spontaneously by HD 3
HD 8	Brain sono: ischemic encephalopathy pattern
HD 15	1st extubation failed → re-intubated. Enteral feeding and PT initiated.
HD 24	Brain MRI: diffuse brain atrophy with ESS; Lt. PVL — high-risk for developmental delay
HD 32	Successful extubation. NIV-NAVA → HFNC → nasal prong → room air (HD 49). Discharged HD 55

Key Investigations

Echocardiography	Huge PDA (6–9 mm, RL shunt), dextroposition, hypoplastic aorta. PDA closed spontaneously by HD 3; PAH confirmed resolved on HD 14
Brain sonography	GMH grade 1 on HD 1. Ischemic encephalopathy pattern on HD 8
Brain MRI (HD 24)	Diffuse brain atrophy with enlarged subarachnoid space (ESS); Lt. PVL — high-risk for developmental delay; rehabilitation initiated
UGIS (HD 29)	No malrotation confirmed; ileus resolved with conservative care
Newborn screening	R/O CPT1A deficiency — acylcarnitine profile and urine organic acid subsequently normal

Discussion

CDH is typically identified at the 18–24 week anomaly scan; however, this case demonstrates that even extensive visceral herniation may escape detection until the third trimester. Retrospective review identified subtle findings as early as 34 weeks, highlighting the importance of serial late-gestation surveillance. The prenatal ultrasound at 39+5 weeks revealed severe cardiac compromise — dextroposition, hypoplastic aortic isthmus, and a massive PDA — and borderline lung volume (O/E LHR 51.4%), underscoring significant pulmonary impact despite late presentation. Prompt EXIT procedure and multidisciplinary NICU care — including aggressive PPHN management with Remodulin as an ECMO-salvage strategy — were key to survival without ECMO.

Key Message

Significant CDH with extensive visceral herniation can remain undetected on routine mid-trimester screening — even when cardiac compromise and pulmonary hypoplasia are present at birth. Late diagnosis does not necessarily portend a poor outcome when prompt referral, EXIT procedure, and coordinated multidisciplinary management are implemented. Sustained vigilance during late gestation and thorough perinatal planning are essential for optimizing neonatal outcomes.