



# EP05.05. When a diaphragmatic hernia is not what it seems: prenatal diagnosis of congenital hiatal hernia mimicking congenital diaphragmatic hernia



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## Background

Congenital hiatal hernia (CHH) is a rare condition that may mimic congenital diaphragmatic hernia (CDH) on prenatal imaging, with diagnosis largely limited to case reports. Accurate differentiation is essential due to differences in prognosis and management.

## Case Presentation

We report two prenatal cases of CHH associated with complex congenital heart disease. The first case was a dichorionic diamniotic twin pregnancy conceived via IVF. At 28+6 weeks, fetus A had conotruncal anomalies, including double outlet right ventricle, ventricular septal defect, and pulmonary stenosis. The second case was a singleton IVF pregnancy with right atrial isomerism, referred at 36+6 weeks.

In both cases, ultrasound showed a retrocardiac cystic structure between the heart and spine, continuous with the stomach, with dynamic movement and no mediastinal shift. No pleural or pericardial effusion was observed. Fetal MRI confirmed CHH. Both neonates underwent cardiac surgery. No intervention was required for CHH. One neonate died postoperatively, while the other remains alive at 19 months with recurrent pneumonia.

## Discussion

CHH generally requires no intervention unless it is giant, whereas CDH carries higher mortality (16.2%, n=121/747) and often requires postnatal treatment. Sonographic differentiation relies on stomach mobility and visualization of an intact diaphragm on a parasagittal view.

## Conclusion

Unlike CDH, which involves a true diaphragmatic defect, CHH represents herniation through the esophageal hiatus and should be considered in the differential diagnosis of retrocardiac thoracic cystic lesions to avoid misdiagnosis and guide management.

## Key message

Isolated CHH has a good prognosis. However, when CHH is associated with complex congenital heart disease, both prognosis and management depend primarily on the cardiac defect.

CHH alone does not warrant pregnancy termination.



Figure 1. Case 1 (28+6 weeks): The four-chamber view (top-right image) revealed a retrocardiac cystic structure located between the heart and the spine, without any mediastinal shift. The parasagittal view (middle) showed the connection of the cystic structure to the stomach at the abdominal level. The fetal MRI (bottom) confirmed herniation through the esophageal hiatus, as indicated by the arrow.



Figure 2. Case 2 (36+6 weeks): The axial view (left) showed a retrocardiac cystic structure located between the heart and spine, with no evidence of mediastinal shift or pericardial effusion. The coronal view (middle) indicated continuity with the stomach, while the fetal MRI (right) confirmed herniation through the esophageal hiatus, marked by the arrow.