CASE SERIES

Congenital Diaphragmatic Hernia: Antenatal Sonographic Findings with Review of Literature

Amandeep Singh[1], Japsimran Kaur[2], Guneet[3]

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Abstract

Background: Congenital diaphragmatic hernia (CDH) is a defect in the diaphragm usually detected on a prenatal scan. The incidence is approximately 1:4,000 live births. The diaphragmatic defect is frequently left and posterolateral (Bochdalek). Surgical repair is possible and easier in neonates; however, the mortality remains high because of pulmonary hypoplasia and pulmonary vascular changes. The likely course of outcome of this condition, largely depends on lung volume on the contralateral side.

Purpose: The aim of antenatal imaging is for early detection. This study presents different cases of CDH along with their imaging features on 2D and 3D ultrasonography. Assessment in tertiary care centers would help in identifying the prognostic factors which aid in planning antenatal management. When a defect is identified by ultrasound before 25 weeks of gestation, a careful ultrasound scan may suggest features that have an isolated diaphragmatic abnormality and a great chance of survival.

Material and methods: About 4 cases were evaluated during the second trimester or anomaly scan over a period of 1 year. The scans were conducted between 24 and 30 weeks of gestation and referred to the Department of Radiodiagnosis and Imaging by the consulting obstetrician.

On imaging the presence of an abdominal organ, most likely the stomach, within the fetal chest directs the diagnosis toward CDH. On four-chamber view of heart, the stomach is seen just behind the left atrium and ventricle in the lower thorax in a left-sided CDH.

Keywords: Bochdalek, Case report, Congenital, Diaphragm, Hernia, Neonate, Pregnancy.

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Case Description

Case 1

A 32-year-old gravida 2 patient with a history of one abortion was referred for antenatal ultrasound with 7-month amenorrhea. Her clinical history was unremarkable but her targeted anomaly scan wasn't done. Her routine nuchal translucency (NT) scan was unremarkable. On ultrasound fetal growth parameters were normal corresponding to a mean gestational age of 26 weeks of gestation; with a normal amniotic fluid index and unremarkable doppler study.

Examination of the fetal chest revealed herniation of the stomach into the thoracic cavity adjacent to the heart (Fig. 1). In the sagittal and axial sections at the level of the thorax in the antenatal ultrasound stomach bubble is seen at the level of the heart in the region of the thorax.

Case 2

A 31-year-old G3 P2 A0 female was referred for the routine antenatal scan at our institute. An old antenatal scan done at 16 weeks of gestation revealed a normal fetus with cardiac activity and all growth parameters corresponding to gestation age. No previous NT-NB and anomaly scan (level II) were available. The ultrasound at our institution was done at 25 weeks and 4 days. Sonographic findings showed a posterior placenta with the increased amount of liquor (AFI-28). The stomach is seen at the same level as the transverse level as that of the heart. On CDFI, normal color within the heart. In these axial sonographic images Figure 2 stomach is seen at same the transverse level as that of four-chamber view of the heart. It is also observed that there is a hypoplasia of the left lung with normal looking right lung suggestive of left-sided congenital diaphragmatic hernia (CDH).

[1–3]Department of Radiodiagnosis, Sri Guru Ram Das Institute of Medical Sciences and Research, Amritsar, Punjab, India

Corresponding Author: Amandeep Singh, Department of Radiodiagnosis, Sri Guru Ram Das Institute of Medical Sciences and Research, Amritsar, Punjab, India, Phone: +91 9872454954, e-mail: dr.amancs@gmail.com

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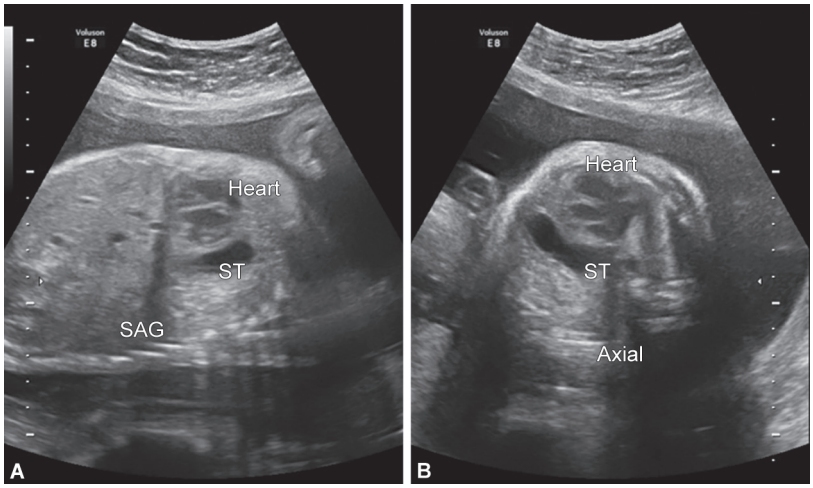
Case 3

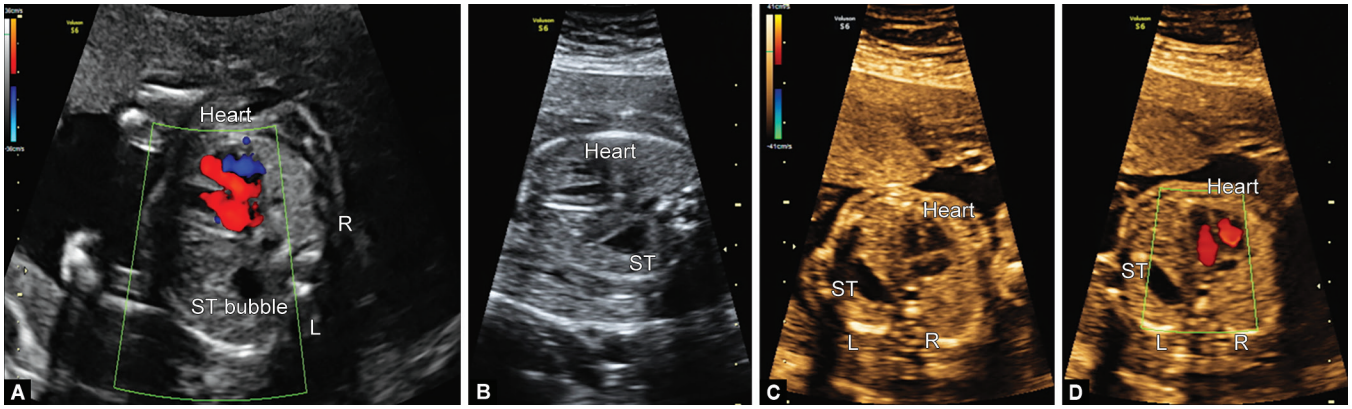
A 25-year-old primigravida was referred for her first antenatal ultrasound at our institute. Antenatal ultrasonogram was done at 28 weeks 5 days gestation according to last menstrual period (LMP). The fetus was in a cephalic position at the time of presentation with the anterior placenta lying 4 cm above the internal OS. Figure 3 in the axial section at the level of thorax, the stomach bubble was seen in the transverse plane at the level of heart and not in the abdomen.

Case 4

A 29-year-old G2 P1 female presented for an antenatal scan at our institute. Previous NT-NB scans done elsewhere revealed no gross congenital anomaly. The present ultrasound was done at 29 weeks according to her LMP. Placenta was anterior and upper with adequate liquor. Figure 4 in the sagittal section at the level of thorax and abdomen—there is reduced abdominal circumference and the stomach is visualized at the same transverse level as the



Figs 1A and B: In the sagittal and axial sections at the level of the thorax in the antenatal ultrasound stomach bubble is seen at the level of the heart in the region of the thorax



Figs 2A to D: In these axial sonographic images, stomach is seen at same the transverse level as that of four-chamber view of the heart. It is also observed that there is a hypoplasia of the left lung with normal looking right lung suggestive of left-sided congenital diaphragmatic hernia

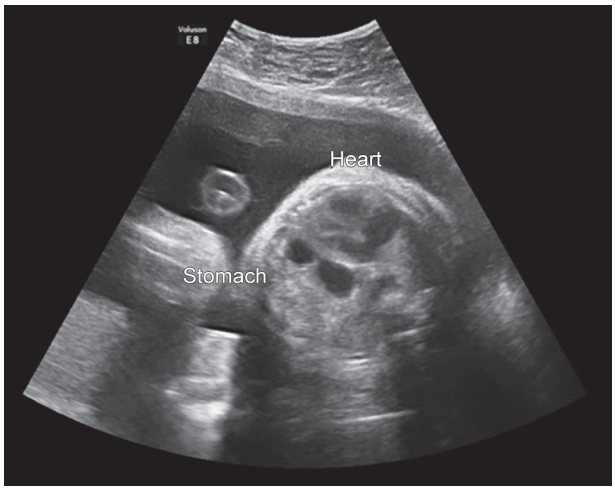


Fig. 3: In the axial section at the level of thorax, there is the absence of a stomach bubble in the abdomen and was seen at the same transverse level as that of the heart

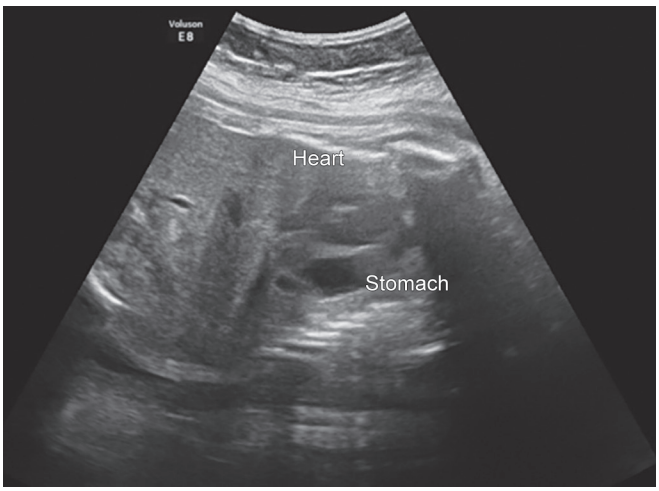


Fig. 4: In the sagittal section at the level of thorax and abdomen—there is reduced abdominal circumference and the stomach is visualized at the same transverse level as the heart, suggestive of congenital diaphragmatic hernia

heart, pointing towards the diagnosis of congenital diaphragmatic hernia.

Discussion

Congenital diaphragmatic hernia is a congenital defect in the diaphragm usually detected on prenatal scans with an approximate incidence of 1:4,000 live births. The diaphragmatic defect is frequently left and posterolateral (Bochdalek). The defect causes the herniation of abdominal contents into the thoracic cavity. At the time of herniation lung development, bronchial branching and development of pulmonary artery happens. As a result, there is a reduction in terminal bronchioles, alveolar septal thickening, and decreased volume with alveoli with accentuation of the thickness of the medial arterial wall. The consequence is reduced surface area for gas exchange, and fixed increase of vascular resistance causing lung compression with resultant pulmonary hypoplasia. The fixed pulmonary and vascular hypoplasia, reversible, pulmonary vascular reactivity causes postnatal compromise with resultant respiratory failure and persistent fetal circulation.[1–5]

Major postnatal sequelae of CDH are chronic lung disease (CLD)/prematurity or bronchopulmonary dysplasia. Chronic lung disease is characterized by damaged and inadequately functioning new-born lung tissue causing breathing and health problems. The symptoms could be due to entrapment of air or collapse of the lung, excessive mucin, or fluid. Children with adequate perinatal treatment are found to have better prognosis with respect to children, not delivered in tertiary care centers. This should serve as a motivation for improvising the prenatal prediction of postnatal outcome in order to, to evaluate if the delivery should be done at a tertiary care center.

Surgical repair is possible and easier in neonates; however, the mortality remains high because of pulmonary hypoplasia and pulmonary vascular changes.

Early diagnosis of CDH is linked with a significant increase (about 2–4 times) in postnatal mortality which may be because larger defects are detected earlier and could have associated anomalies. The mean age of diagnosis is around 24 weeks. No association is found in approximately 50% of cases which survival is based on the volume of the contralateral lung. About 25% of cases are associated with chromosomal anomalies and the rest of the 25% of cases are linked with other anomalies or syndromes. Bochdalek hernia which is a posterolateral defect constitutes around ninety percent of cases.[6] This defect on the left side leads to herniation of the left liver lobe, bowel, spleen, and stomach into the ipsilateral chest with contralateral mediastinal shift, eventually leading to pulmonary hypoplasia and pulmonary hypertension. The outcome is based on various factors like early diagnosis, contralateral lung volume, cardiac function, associated anomalies, pulmonary vasculature, and gestational age at which delivery occurs. Congenital diaphragmatic hernia is usually isolated but could be associated with trisomies 18 or 13 and tetrasomy 12p or Pallister–Killian syndrome.[7]

In our study only isolated CDH was present and was not associated with any other syndrome was found.

The lung area to head circumference ratio (LHR), the observed/expected lung-to-head ratio (o/e LHR), and the quantitative lung index (QLI) are used to calculate the prognosis in case of CDH.[8,9]

Quintero and colleagues have proposed, that the observed/expected LHR and the LHR value were not independent of gestational age mathematically and caution should be kept when using them in predicting outcome. The ratio between the contralateral lung area taken at the level of four-chamber view of the heart and the circumference of the fetal heart is the LHR. It determines the severity of hypoplasis of lung. Lung–to–head ratio values less than 1 are associated with very low survival rates with values between 1–1.4 and more than 1.4 associated with moderate and high survival rates respectively. In this case, LHR was less than 1 with an observed-to-expected LHR percentage (O/E LHR) of 24% and was thus having a bad prognosis.[10–14]

The aim of our study in prenatal imaging is to establish an early diagnosis. This study presents different cases of CDH along with their imaging features on 2D and 3D ultrasonography. Assessment in tertiary care centers would help in identifying the prognostic factors which aid in planning antenatal management. When a defect is identified by ultrasound before 25 weeks of gestation, a careful ultrasound scan may suggest features that have an isolated diaphragmatic abnormality and a great chance of survival. Four cases were evaluated during the second trimester or anomaly scan over a period of one year.

Visualization of the abdominal organ, most likely the stomach, within the fetal chest, directs the diagnosis towards CDH. On four-chamber axial view of heat the stomach is seen just behind the left atrium and ventricle in the lower thorax in a left-sided CDH as seen in Figures 1 to 4.

Conclusion

Congenital diaphragmatic hernia on two-dimensional and three-dimensional ultrasonography.

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