Prenatal Ultrasonographic Diagnosis of Congenital Diaphragmatic Hernia at 11 Weeks Gestation

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Congenital diaphragmatic hernia is a major malformation. Early diagnosis is in itself an important factor for the prediction of the outcome. The authors present a case of left-sided fetal diaphragmatic hernia diagnosed by transvaginal ultrasound examination at 10 weeks, 5 days gestation.

Key words: diaphragmatic hernia, prenatal diagnosis, prenatal ultrasonography, Doppler ultrasound, transvaginal ultrasonography

One of the most important roles of prenatal ultrasonography is to detect fetal malformations as early as possible. Early diagnosis makes the termination of the pregnancy possible with less complication, and there is time for planning for further follow-up and interventions.[1]

Congenital diaphragmatic hernia is the fourth most common structural abnormality encountered in the neonate.[2] The prenatal diagnosis of diaphragmatic hernia by ultrasonography usually is made during the second trimester of pregnancy by the demonstration of intrathoracic viscera and the associated mediastinal shift.[3] With the advent of vaginal scanning that permits high-frequency, high-resolution ultrasound imaging, the diagnosis is increasingly made in the first trimester.[4,5] To the best of our knowledge, the earliest ultrasound diagnosis of congenital diaphragmatic hernia reported in the literature was made at 12 weeks gestation.[6] We report transvaginal sonographic findings of congenital diaphragmatic hernia diagnosed at 10 weeks, 5 days gestation.

Case Presentation

A 29-year-old woman, gravida 3, para 0, had a transvaginal ultrasound examination using a LOGIQ Pro 200 ultrasound machine (General Electric, Wakasee, WI) with a 5-MHz transvaginal transducer at 11 weeks gestation for dating. It showed a single fetus with a crown-rump length of 39 mm that is consistent with 10 weeks, 5 days gestation. The nuchal

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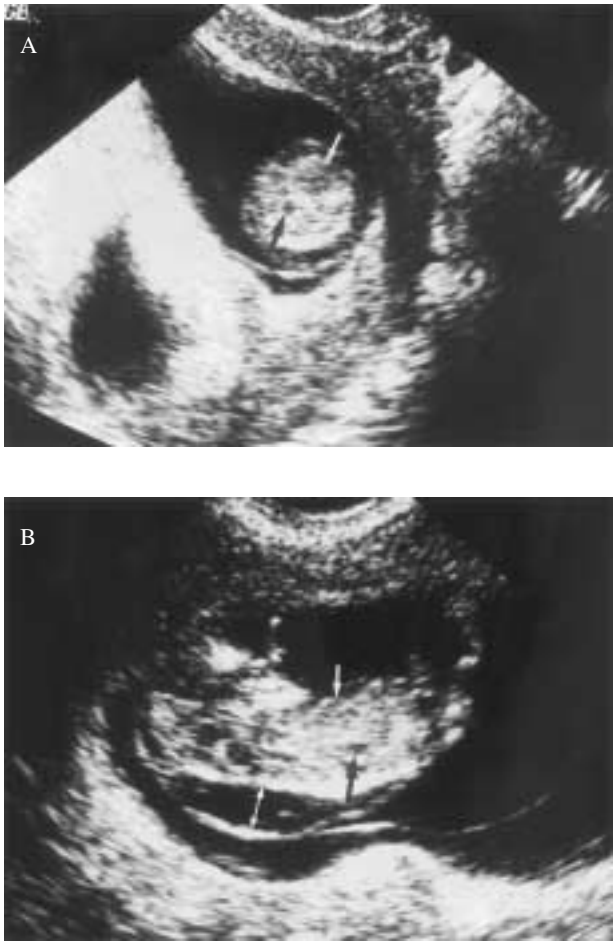

FIG. 1. Transvaginal scan at 10 weeks, 5 days gestation. (**A**) Axial image of the fetal chest shows the heart (small arrow) in right side and the stomach (large arrow) in left aspect to the heart at the same plane, (**B**) oblique sagittal image shows the elevated stomach (large arrow) at the level of the heart (small arrow). The thickness of nuchal translucency (bidirectional arrow) is about 5 mm.

translucency was 5 mm. The fetal heart was located in the right chest, and the stomach was seen in left aspect to the heart at the same level (Fig. 1). Left-sided diaphragmatic hernia associated with hydrops fetalis or cystic hygroma was suspected. Follow-up ultrasound with color Doppler imaging was performed at 12 weeks gestation. The fetal heart and the elevated stomach were well distinguished from each other (Fig. 2). Early amniocentesis at 14 weeks gestation revealed a normal karyotype. At 18 weeks gestation, left-sided diaphragmatic hernia was confirmed by targeted ultrasonography using a LOGIQ 700 magnetic resonance

FIG. 2. Color Doppler image at 12 weeks shows flow signals in the heart. H = heart, ST = stomach.

ultrasound machine (Fig. 3). A single umbilical artery was also noted.

Because a congenital diaphragmatic hernia detected at such an early stage of pregnancy is usually considered to have a poor clinical outcome, termination of pregnancy was performed at 18 weeks gestation. The autopsy of a 200-g male fetus showed left-sided diaphragmatic hernia, Meckel's diverticulum, and single umbilical artery.

Discussion

Diaphragmatic hernia can be diagnosed by the ultrasonographic demonstration of stomach, intestines, or liver in the thorax and the associated

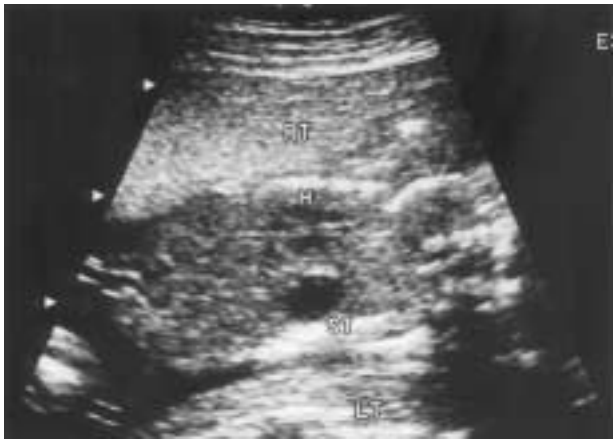

FIG. 3. Transabdominal scan at 18 weeks gestation. On the coronal image, the stomach is located in the left side of the chest and the heart is displaced to the right side. H = heart, ST = stomach.

mediastinal shift to the opposite side. In about 50% of affected fetuses, there are associated chromosomal abnormalities or other defects.[7] Although congenital diaphragmatic hernia is an anatomically simple and easily correctable defect, the reported mortality rate ranges between 24% and 95%,[8] which in the absence of additional malformations is due mostly to respiratory insufficiency secondary to pulmonary hypoplasia and persistent pulmonary hypertension. Intrathoracic compression of the lungs by the herniated abdominal viscera and alterations of the pulmonary blood flow appear to be major factors in the pathogenesis of the pulmonary hypoplasia in congenital diaphragmatic hernia.[9] Intrauterine surgery may be performed before 24 weeks gestation because important fetal lung development occurs in the mid second trimester. Early ultrasound recognition is therefore necessary.[10]

Color Doppler ultrasonography was very helpful in the diagnosis of our case. Because the fetal stomach is not large enough to confirm at such an early gestational age, definite identification of fetal stomach and heart may be unavailable with gray-scale imaging. After applying the color Doppler to the image, the fetal stomach and heart were well distinguished from each other.

The nuchal translucency was increased in this fetus. The diaphragm develops by fusion of the pleuroperitoneal membrane with the septum transversum, the dorsal mesentery of the esophagus, and the lateral body wall. This process usually is completed by 9 weeks gestation.[11] In the presence of a defective diaphragm, there is herniation of the abdominal viscera into the thorax at about 10 weeks gestation, when the intestines return to the abdominal cavity from the umbilical cord. It is possible that in those cases with increased nuchal translucency, there is intrathoracic herniation of the abdominal viscera at this gestational age. Increased nuchal translucency may be the consequence of venous congestion in the head and neck due to mediastinal compression and impedance of venous return. In such cases, prolonged intrathoracic compression of the lungs causes pulmonary hypoplasia.[3]

First-trimester fetal malformation screening still represents a diagnostic challenge in modern obstetrics, being solely dependent on ultrasound instrumentation and on the skill and experience of the operator, recently improved by the introduction of the high-frequency transvaginal approach and digital image processing. As detail resolution improves, combined with knowledge of the normal embryological development and its corresponding ultrasound findings, the potential for first-trimester malformation screening is enhanced.[12]

Quashie et al.[13] investigated the ability to visualize the first-trimester fetus by transvaginal sonography with increasing gestational age (8 to 13 weeks) in 60 women. It was suggested that by 12 to 13 weeks gestation, most of the fetal organs can be visualized in similar detail to that required to complete a second-trimester anomaly scan.

Whitlow et al.[14] reported that the detection rate of structurally abnormal fetuses was 59% in early pregnancy and 81% in combination with the second-trimester scan. A broad range of fetal abnormalities were diagnosed in the first trimester, with 64.7% sensitivity for the detection of major structural abnormalities.[15] The commonest anomalies detected by transvaginal ultrasonography in the first trimester are anomalies of the head and brain, heart, abdominal wall, umbilical cord, urinary tract, and skeleton.[12]

Therefore, not only measurements of fetal size but also detailed anatomical evaluation for the fetal major organs should be included in first-trimester ultrasonography. In the transvaginal ultrasonography performed at 10 to 14 weeks gestation, we always evaluate the fetal major organs such as skull shape and falx, posterior neck (for measuring nuchal translucency), heart, stomach, kidneys, bladder, abdominal cord insertion site, and extremities.

The high-resolution transvaginal sonography combined with color Doppler sonography is very useful in detecting major fetal structural abnormalities such as congenital diaphragmatic hernia, in this case in the first trimester. Thorough anatomical evaluation for fetal major organs at 10 to 14 weeks gestation is recommended for early detection of major fetal structural abnormalities.

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