

# *First-Trimester Ectopia Cordis in a Twin Gestation, Using 3D Surface Rendering and Reconstruction*

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Ectopia cordis is a very rare and impressive congenital abnormality. This case study demonstrates an 8-week, 5-day twin gestation with one embryo affected and the other normal. The abnormal fetus spontaneously expired at 11 weeks, 5 days gestation. The surviving twin remains alive and well at 18 weeks gestation. Because of the ongoing normal pregnancy, pathologic confirmation is unlikely.

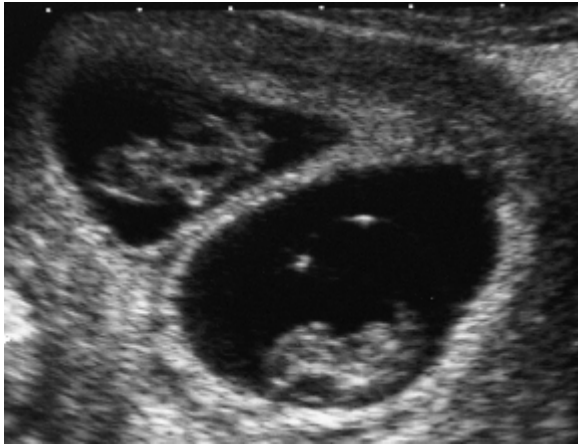
*Key words:* ectopia cordis, pentalogy of Cantrell, 3D sonography

Ectopia cordis is a rare malformation of the ventral wall resulting in the partial or total evisceration of the heart through the defect. It occurs in 5.5 to 7.9 per million live births.<sup>1-7</sup> Ectopia cordis has a very poor prognosis. Few fetuses with this anomaly will reach delivery. There is a high mortality rate prenatally and in the neonatal period. With advances in surgical techniques some postpartum infants have been successfully treated and have survived.

## **Case Report**

A gravida 4, para 1 patient in her early 40s presented for a first-trimester examination for uncertain dates. A transvaginal examination using a Toshiba Nemio sonographic system, with a phased-array 6.5-MHz transducer, revealed a diamniotic, dichorionic, 6-week, 2-day twin intrauterine gestation. Two small embryos were identified with normal heart rates. The patient returned approximately 2 weeks later for anxiety and reassurance. During a transabdominal examination, one embryo appeared normal, whereas the other appeared to have a very prominent heart (Fig. 1). Transvaginal examination revealed the embryonic heart protruding outside the thorax, with fluid occupying both lung fields (Figs. 2, 3). Multiple 3D surface renderings were obtained using Bio-

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**FIG. 1.** Twin gestation with one normal (lower) and one abnormal (upper) embryo.

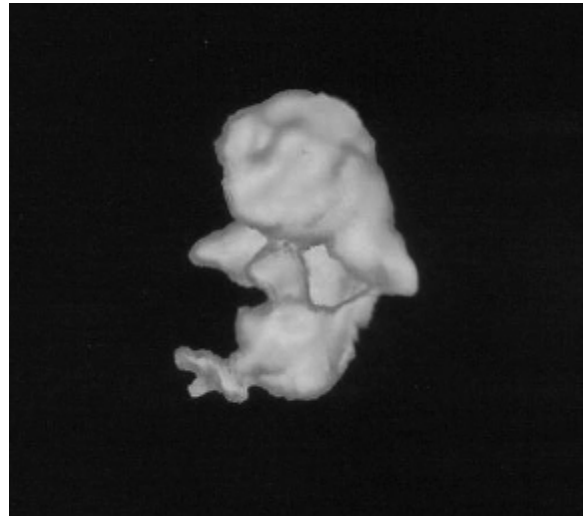


**FIG. 2.** Abnormal embryo with the heart extending through the thoracic defect.

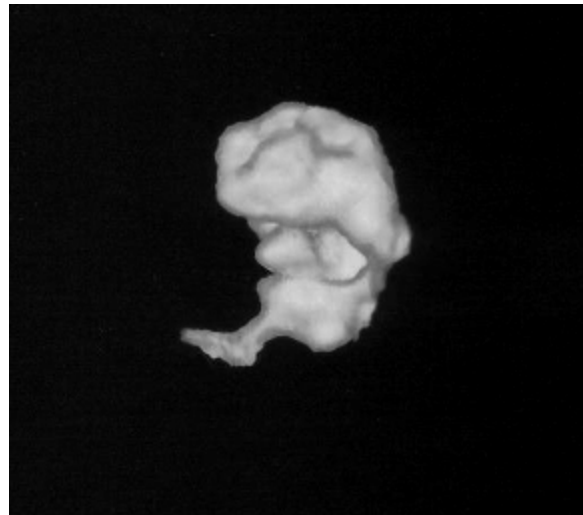


**FIG. 3.** Another image of the abnormal embryo with the heart extending through the thoracic defect.

medicom Sonreal/Sono 3D software. While the diagnosis was reasonably obvious with the 2D

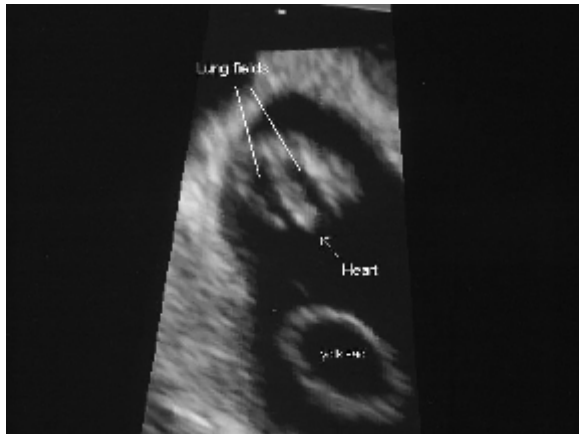


**FIG. 4.** One 3D surface rendering of the abnormal embryo.



**FIG. 5.** Another 3D surface rendering of the abnormal embryo.

examination, the 3D images delineated the abnormality in a dramatic fashion, making the diagnosis undeniable (Figs. 4, 5). The heart is seen extending well beyond the plane of the chest wall. Further image processing using multiplanar reconstruction in a transverse plane provided superb detail of the protruding heart tethered posteriorly, extending through a large thoracic defect, with both lung fields completely fluid filled (Fig. 6). An examination at 11 weeks, 5 days at another facility demonstrated spontaneous demise of the abnormal fetus. The surviving twin is alive and well at approximately 18 weeks gestation. Because of the ongoing

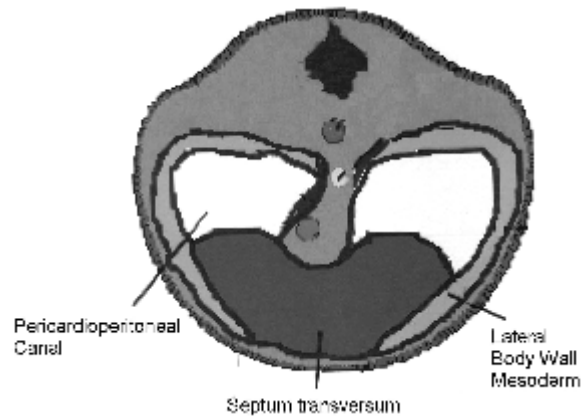


**FIG. 6.** Transverse reconstruction of the abnormal thorax demonstrating fluid-filled lung fields.

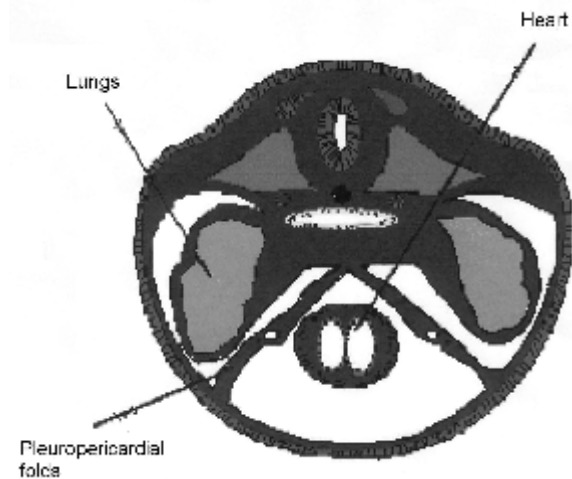
normal pregnancy, pathologic confirmation is unlikely.

### Discussion

During the third week of gestational life, when the demarcation between the intraembryonic and extraembryonic coeloms is established, development of the body cavities and diaphragms begins. By the beginning of the fourth week, a horseshoe-shaped cavity develops in the cardiogenic and lateral mesoderms (Fig. 7). The curve of the horseshoe represents the future pericardial cavity. During this period, the pericardial and peritoneal cavities communicate dorsally through the pericardioperitoneal canals. Caudally, the developing septum transversum will ultimately separate the thoracic and abdominal cavities by formation of the diaphragms. The fibrous pericardium forms from the pleuropericardial folds. The folds grow toward the midline and attach to the primitive mediastinum and end up separating the heart from the lungs, with the thoracic cavity now divided into a pericardial cavity and two pleural cavities (Fig. 8). During folding of the embryonic disc in week 4, the lateral body walls converge together on the ventral aspect of the embryo and fuse.<sup>1-7</sup> It is at this stage that incomplete fusion may occur and result in the partial or complete evisceration of the heart through the defect. The etiology of this process is unknown. The embryological development of this anomaly may be related to early fetal rupture of the



**FIG. 7.** Body cavity development showing early pericardial cavity.



**FIG. 8.** Body cavity development showing formation of pleuropericardial folds.

amnion, chorion, or yolk sac, as well as teratogens, or chromosomal aberrations.<sup>7,8</sup>

Ectopia cordis is a very rare malformation of the ventral wall resulting in the partial or total evisceration of the heart through the defect. It occurs in 5.5 to 7.9 per million live births. It is generally divided into five types: cervical, cervicothoracic, thoracic, abdominal, and thoracoabdominal. The two most common forms are thoracic (59%) and thoracoabdominal (38%). In many cases, the thoracoabdominal type is characterized by partial absence or cleft of the sternum, deficiency of the anterior diaphragm, deficiency of the diaphragmatic pericardium, midline abdominal wall defect, and

congenital intracardiac abnormalities. These five characteristics comprise the pentalogy of Cantrell.<sup>1-7</sup>

Ectopia cordis carries a poor prognosis, particularly in the presence of the thoracic type, with significant heart defects. Most fetuses with this anomaly will never reach delivery, and those that do deliver have a very high mortality rate during the neonatal period. With advances in surgical techniques increasing, small numbers of postpartum infants have been successfully treated and have survived. The goal of the initial management is directed at providing coverage of the bare heart with skin or synthetic material before infection can significantly compromise survival. Subsequent operations to reconstruct the chest cavity include enlarging the thoracic cage by shifting the diaphragmatic attachments and moving the heart into an intrathoracic position. This may be difficult because of kinking of the great vessels.

Sternal reconstruction usually is required later on, using a variety of techniques, including use of native materials (rib or sternal transfer) and/or insertion of prosthetic devices.<sup>9-12</sup>

### Conclusion

The prenatal diagnosis of ectopia cordis in the first trimester is uncommon. This case report provides an excellent opportunity to review the possible origins, development, prenatal diagnosis, and

treatment of this rare and exotic embryologic anomaly.

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