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CASE REPORTS

Ectopia Cordis: Case of an Early Diagnosis and Review of Literature

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Introduction

Ectopia cordis is a severe congenital condition in which the heart is completely or partially displaced outside of the thoracic cavity [1–3]. The term was derived from the Greek word *ektopos* meaning away from a place. This anomaly was first described by Stensen in 1671 [4]. Byron [5] classified ectopia cordis into four categories: cervical, thoracic, thoracoabdominal, and abdominal depending on the location of the ectopic heart.

The thoracoabdominal group has been described as a separate syndrome of five associated anomalies: (1) a midline supraumbilical abdominal wall defect, (2) a defect of the lower sternum, (3) a deficiency of the anterior diaphragm, (4) a defect in the diaphragmatic pericardium, and (5) congenital intracardiac defects [6]. With few exceptions, the outcome is poor because of intracardiac and extracardiac structural abnormalities that are almost always present [1–3].

Several researchers have documented a pulsating heart that is partially or totally outside the thoracic cage as a prenatal diagnosis of ectopia cordis [1, 3]. In most cases, the diagnosis is simple and can be done with confidence as early as 10 weeks of pregnancy [7, 8]. If only the apex of the heart is extra-thoracic and visibility is hampered by extruded abdominal material, cases of thoracoabdominal ectopia cordis may be difficult to diagnose. With the widespread use of first trimester sonography as a screening technique for aneuploidy in modern clinical practice, it is reasonable to assume that the majority of cases of ectopia cordis will be detected. This report aims to document one such case of ectopia cordis which was diagnosed as early as 7 weeks of gestation.

Case Review

A primigravida in her late 20s presented for a first-trimester examination for uncertain dates. A transvaginal examination using a GE VOLUSON S8 CORE sonographic system revealed a single live, 7 weeks, 3 days intrauterine gestation. An embryo was identified with the presence of cardiac activity. Transvaginal examination revealed the embryonic heart protruding outside the thorax. The diagnosis was reasonably obvious with still images as well (Fig. 1); videos delineated the abnormality in a dramatic fashion, making the diagnosis undeniable. The heart was seen extending well beyond the plane of the chest wall. The lady returned after 10 days with a spontaneous miscarriage at home. Products of conception were not available for further testing.

Discussion

Ectopia cordis is estimated to affect 0.7–0.8 out of every 100,000 deliveries [2]. Ectopia cordis is thought to be the result of a halt in the folding process of the heart which

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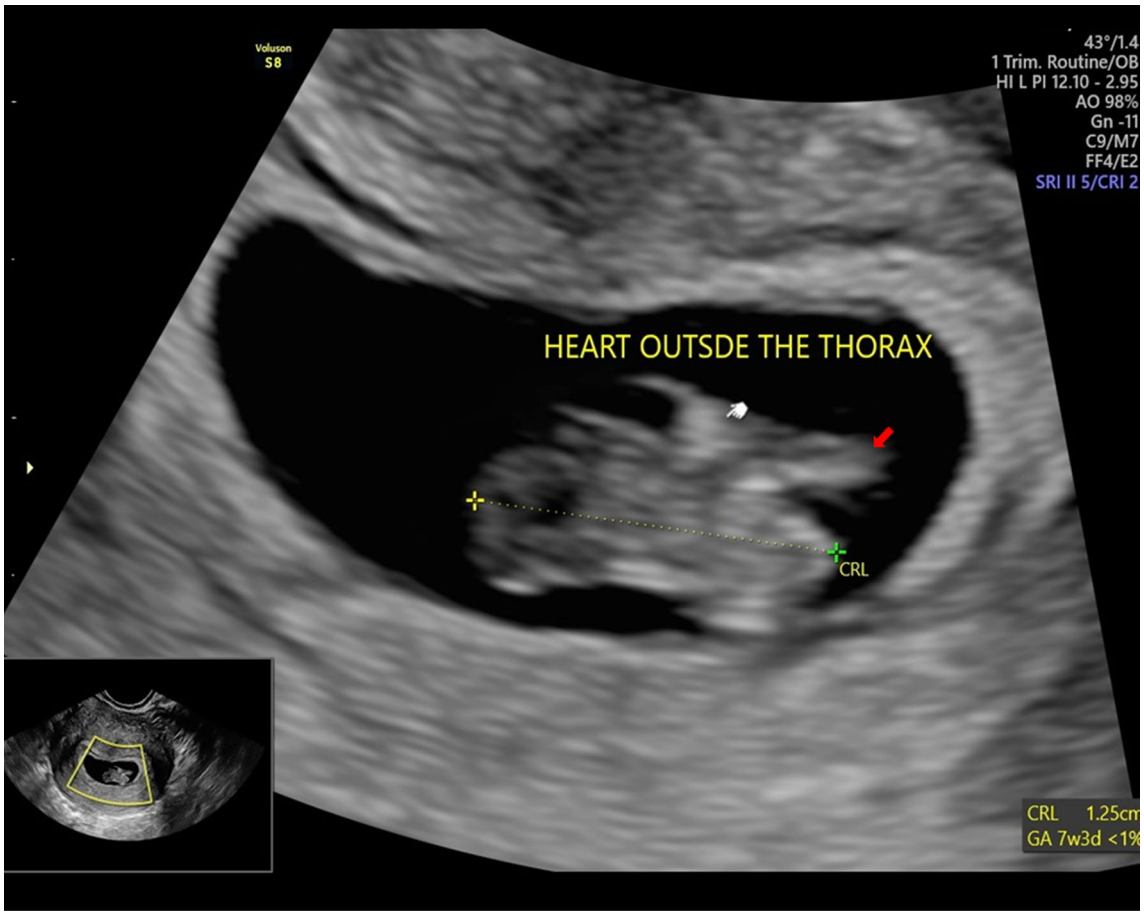

Fig. 1 Echogenic myocardial walls (white arrow) are seen protruding outside the fetal body plane. This is associated with other contents (red arrow), diagnosis of which cannot be made with certainity at this gestation

normally occurs after the fourth week of development [9]. The heart is located outside the body throughout early stages of development. The heart passively moves into its final intrathoracic location and gets included in the pericardial sac when the embryo folds (Fig. 2). Although the physiologic mechanisms that lead to ectopia cordis are unknown, mechanical distortion of the developing heart caused by early rupture of the chorion and/or yolk sac could be a contributing factor [10].

Ectopia cordis is commonly diagnosed prenatally with sonography [1, 3]. The first case was diagnosed in a 34-week fetus in 1981 [11] and first trimester diagnosis with transvaginal sonography was achieved a decade later [12]. A high rate of intracardiac and extracardiac abnormalities has been documented in prenatal research.

Although ectopia cordis is generally perceived as a sporadic disorder, a few occurrences have been linked to aneuploidy [13, 14], ectopia cordis is generally associated with anterior abdominal wall defects [15]. Omphalocele can be easily delineated on a first trimester scan and there is a strong link to chromosomal abnormalities, particularly trisomy 18. Some occurrences of Cantrell pentalogy with omphalocele in the first trimester have been linked to ectopia cordis [16].

Ectopia Cordis has a bad prognosis, especially when the thoracic variety is present and with major cardiac abnormalities. The majority of fetuses with this defect never make it to delivery and those that do have a significant neonatal mortality rate. Small numbers of postpartum newborns have been successfully treated and have survived as surgical procedures have improved. The goal of early management is to cover the naked heart with skin or synthetic material before infection poses a substantial threat to survival. Enlarging the thoracic cage by repositioning the diaphragmatic attachments and placing the heart into an intrathoracic location are among the subsequent operations to reconstruct the chest cavity. Because of the kinking of the large vessels, this may be a challenge [17].

Diagnosis of ectopia cordis can be easily made on first trimester screening and the earlier the better. But how early is early? The aim of this article is to sensitise workers to routine early pregnancy scan diagnosis. If a crown and a rump are identified and a pulsating heart is seen outside the CRL

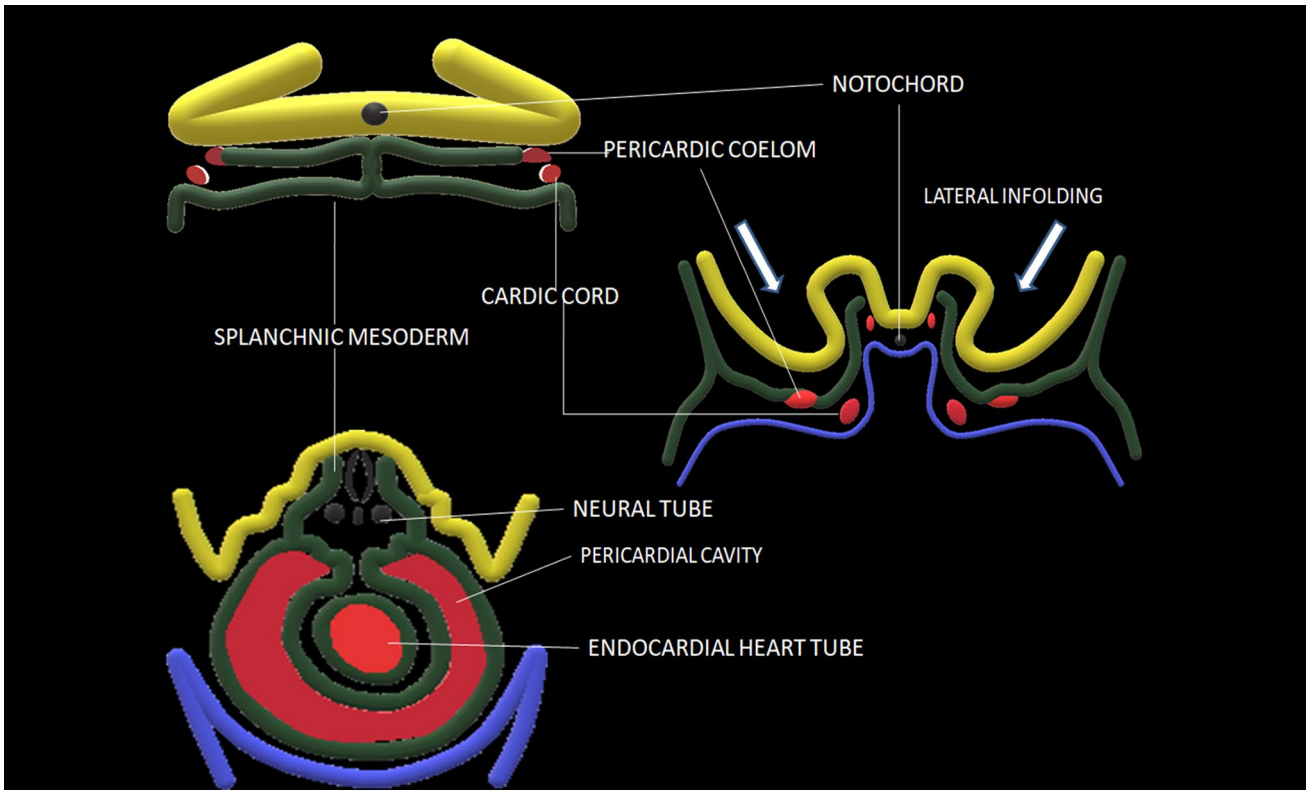

Fig. 2 Step wise lateral infolding of splanchnic mesoderm leads to intrathoracic placement of the heart. During folding of the embryonic disc in week 4, the lateral body walls converge together on the ventral aspect of the embryo and fuse. It is at this stage that incomplete fusion may occur resulting in partial or complete displacement of the heart through the defect

or the body stalk, a suspicion should be made. To the best of our knowledge and extensive literature search, we did not encounter any ectopia cordis diagnosed on ultrasound before 10 weeks of gestation on ultrasound. Given the poor outcome a justifiable option of termination of pregnancy in such cases, early detection could offer an easier and less challenging experience for couples undergoing termination of pregnancy.

Conclusion

Prenatal diagnosis of EC during the first trimester is possible. A growing number of congenital defects are potentially detected at earlier gestational ages as first trimester ultrasonography becomes more widely available and used, and image quality improves. Early diagnosis of EC is desirable to limit physical and psychological trauma and to ensure termination of pregnancy with lower complication rates and shorter hospital stay.

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