Prenatal Diagnosis of Ectopia Cordis

Gokhan Yildirim, Halil Aslan, Kemal Gungorduk*, Ahmet Gul, Emine Unlu
Maternal and Fetal Unit, Bakirkoy Women and Children Hospital, Istanbul, Turkey.

Ectopia cordis is defined as an anomaly in which the fetal heart lies outside the thoracic cavity. It is a rare congenital abnormality with an incidence of 5.5–7.9 per 1 million live births [1]. Ectopia cordis may occur as an isolated malformation or it may be associated with other ventral body wall defects affecting the thorax, abdomen or both. The cause of ectopia cordis is currently unknown, and most cases are sporadic [1,2]. The prognosis is poor, and most infants are stillborn or die within the first few hours or days of life. Attempts at surgical correction have been largely unsuccessful owing to the extent of the associated anomalies. As a result, when ectopia cordis is diagnosed, pregnancy termination before viability or non-aggressive management in the third trimester should be considered and discussed with the parents.

This case report describes the sonographic diagnosis of ectopia cordis in a fetus at 17 weeks' gestation.

A 28-year-old primigravid woman was referred to our maternal and fetal unit for detailed ultrasonography at 17 weeks' gestation. Her husband was 34 years old. The patient had received no regular prenatal care and had no family history of congenital anomalies. She had taken no medication during her pregnancy. Ultrasonographic examination showed a singleton fetus with normal amniotic fluid volume. Fetal biometry was consistent with dates. Detailed ultrasonography revealed a heart situated outside the thoracic cavity (Figure 1). No abdominal, craniofacial or intracardiac abnormalities were detected, and the cardiac outflow tracts appeared normal.

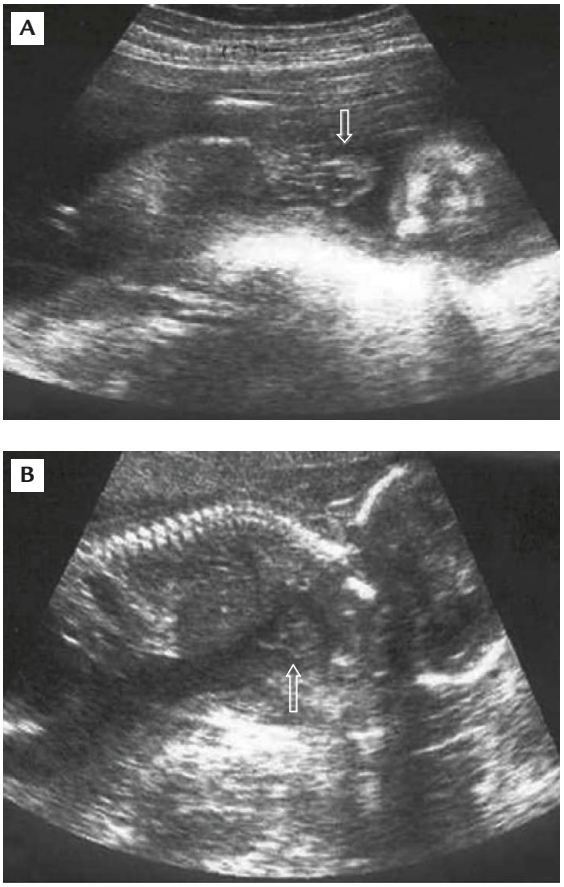

Figure 1. (A, B) Transabdominal sagittal sonogram shows the fetal heart in its pericardial sac, lying completely outside the chest cavity (arrow).

These findings were strongly suggestive of ectopia cordis. The parents were informed of the poor prognosis. Genetic amniocentesis was performed and revealed a normal 46,XY karyotype.

The parents opted for termination of the pregnancy. The mother aborted a 150-g male infant, without complications. Postnatal examination showed that the heart of the fetus was lying outside the thoracic cavity and was devoid of pericardium (Figure 2). The placenta and umbilical cord were unremarkable. The parents refused autopsy.

Ectopia cordis is defined as the complete or partial displacement of the heart outside the thoracic cavity. It is a rare congenital defect in fusion of the anterior chest wall, resulting in an extrathoracic location of the heart. Ectopia cordis is frequently associated with other congenital defects involving multiple organ systems. Ventricular septal defects and tetralogy of Fallot are the most common associated intracardiac defects, while omphalocele is the most common associated abdominal wall defect. In animal models, isolated ectopia cordis

*Correspondence to: Dr Kemal Gungorduk, Maternal and Fetal Unit, Bakirkoy Women and Children Hospital, Yenimahalle, Bakırköy, Istanbul, Turkey.
E-mail: maidenkemal@yahoo.com
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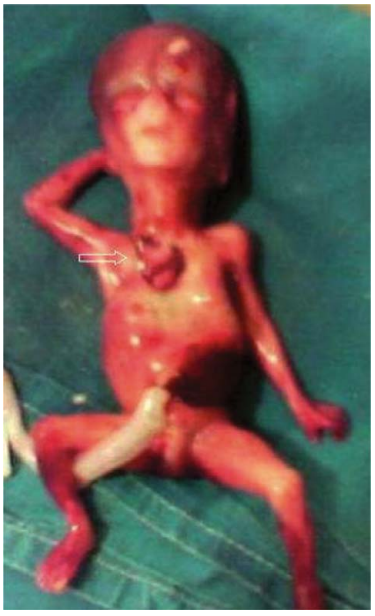

Figure 2. Photograph of the fetus shows ectopia cordis (arrow).

has been attributed to intrauterine drug exposure, a finding which has not been confirmed in humans [1–3]. In our case, no abdominal, craniofacial or intracardiac abnormalities were detected, and the cardiac outflow tracts appeared normal.

The pathogenesis of ectopia cordis and coexisting anomalies has been the subject of research, and there are many theories that attempt to explain this anomaly, including the amniotic band theory, the vascular disruption theory, the theory of a defect in the fetal folding process and the theory of disturbances of field development. Developmental fields are those units of the embryo in which the development of a particular complex structure is determined and controlled in a coordinated, temporally synchronous and hierarchical manner [4,5].

The prenatal diagnosis of ectopia cordis is easily made with ultrasound, which allows visualization of the heart outside the thoracic cavity. Differential diagnoses include limb–body wall complex, amniotic band syndrome, and pentalogy of Cantrell [6]. Prenatal diagnosis of ectopia cordis was reported by Bick et al [8] and Tongsong et al [7] at weeks 11 and 9 of gestation, respectively. In our case, prenatal diagnosis was made at 17 weeks' gestation. While ectopia cordis is generally considered to be an isolated, sporadic malformation, there have been a number of reports linking it to chromosomal abnormalities. Reported karyotype abnormalities include trisomy 18, Turner syndrome and 46,XX,17q+ [3,4]. Our case revealed a 46,XY karyotype. Chromosomal analysis is generally indicated in a patient with prenatally diagnosed ectopia cordis, especially if other anomalies are also identified.

The prognosis is generally poor and depends on the severity of the intracardiac malformations and the presence of associated abnormalities. Most infants are stillborn or die within the first few hours or days of life. Attempts at surgical correction have been largely unsuccessful because of the extent of the associated anomalies.

Obstetric management should include a careful search for associated anomalies, especially cardiac anomalies, and assessment of fetal karyotype. Pregnancy termination prior to viability or non-aggressive management in the third trimester should be considered and discussed with the parents. As this is considered to be a sporadic event, the recurrence risk is not increased over that of the general population [2–9].

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