CASE REPORT

Ectopia Cordis with Associated Foetal Anomalies: Antenatal Ultrasonographic Detection

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Abstract

A rare case of ectopia cordis with unusual associated anomalies detected during antenatal ultrasonographic examination is presented.

Key Words

Ectopic cordis, anomaly.

Introduction

Ectopia cordis is a rare heart lesion that results from an abnormal development of the primitive heart outside the embryonic disc in the early stage of development. It represents a form of pericardial defect but is further characterized by a partial or complete displacement of the heart outside the thorax. Congenital heart diseases like tetralogy of Fallot (most common), venticular hypoplasia, transposition of great vessels (TGA), tricuspid atresia, Ebstein's anomaly, common atrium, ASD, VSD and double outlet right ventricle may be present. Multiple extra-cardiac defects have also been reported with predominence of prolapse of the forebrain, meningocele, encephalocele, cleft lip, palate deformities and ventral wall defects. The prognosis is very poor in these cases.

Case Report

A 24 year old primigravida reported for antenatal examination. Her duration of pregnancy was about 8 months. General physical examination did not show any significant abnormality. On per abdomen examination, head of the foetus could not be well defined. Her routine investigations were within normal limits. She was subjected to ultrasound examination with a suspected diagnosis of anencephaly. Ultrasonography examination revealed that there was a single foetus in the utrine cavity. This was of 32 weeks gestational maturity with multiple anomalies. Cranial vault and brain were missing (Anencephaly). A large irregular cystic mass with thick echogenic septations was seen at cranial pole. A small omphalocele was seen as a midline abdominal wall defect with herniation of liver into the base of umblical cord (Fig. 1). Thoracic cavity was small in size and the pulsatile heart was seen to lie anteriorly on the outer surface of thorax surrounded by amniotic fluid. The great vessels were seen to enter the thoracic

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cavity from the externally placed heart. No gross structural abnormality was detected in the ectopically placed heart (Fig. 2,3).

Patient was explained the poor prognosis carried by the foetus in view of the nature of multiple anomalies. Both husband and wife opted for the termination of pregnancy.

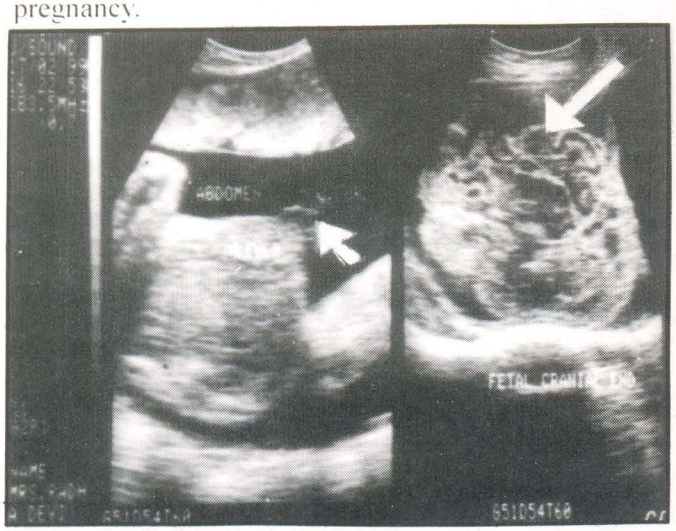

Fig. 1 : Sonogram showing large irregular cystic mass with thick echogenic septation at the cranial end of the foetus (Big Arrow). Vault and brain of the foetus is absent. Small omphalocele with herniated liver at the base of umblical cord (Small Arrow).

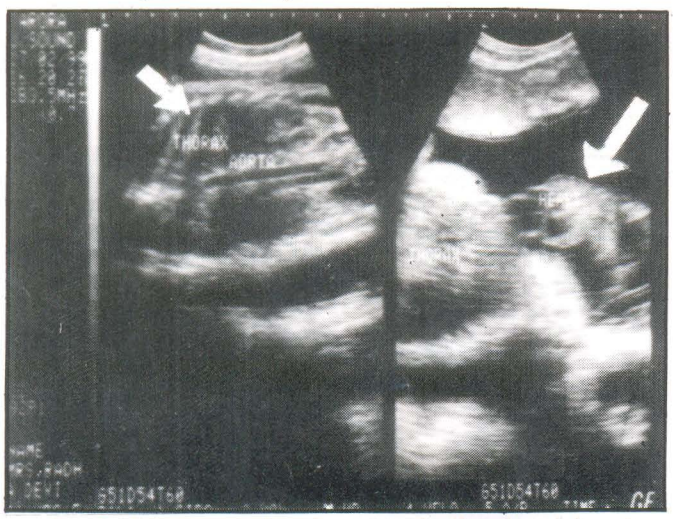

Fig. 2 : Sonogram showing ectopically placed heart outside the thoracic cavity (Big Arrow) and small thoracic cavity (Small Arrow).

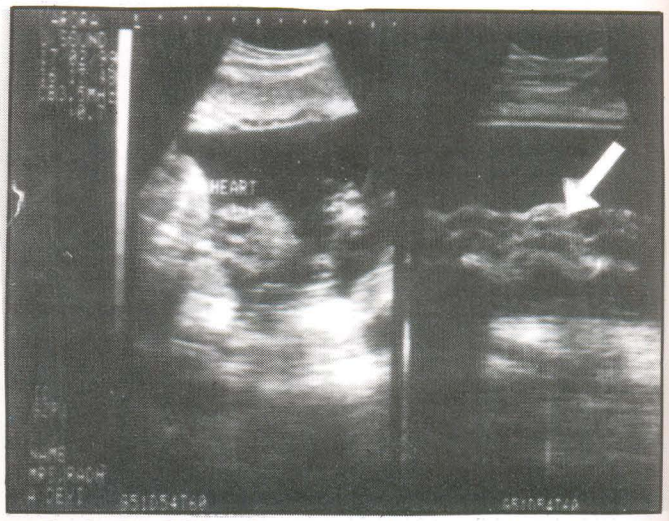

Fig. 3 : The B/M mode sonogram showing cardiac activity of the ectopic heart (Arrow).

Discussion

Abbot qualified ectopia cordis as a "displacement so that the heart passes out of the thorax and comes to lie either on the outer surface of the body or in the abdominal cavity" (1). Stenson first reported a case of ectopia cordis associated with tetralogy of Fallot in 1671 (2). Kanagasuntheram and Verzin suggested a classification including five types : Cervical, throacocervical, thoracic, thoracoabdominal, and abdominal (3). Van Praagh *et. al.* classified ectopia cordis as represented by four types– Cervical, thoracic, thoracoabdominal, and abdominal–but suggested that for practical purposes only two types, thoracic and thoraco-abdominal, are clearly represented clinically (4).

Cervical forms are rare and may simply represent retention of the heart in its embryonic position in the neck.

Thoracic type is the classical form of ectopia cordis as is seen in our case. It is characterized by a sternal cleft that allows protrusion of the heart outside the chest cavity. Complete absence of the parietal pericardium, cephalic orientation of the cardiac apex, epigastric omphalocele or diastasis recti, and a small thoracic cavity.

The throracic type occurred in 80 cases (37%) in the review by Leca *et. al.* (5) Congenital heart disease, most commonly tetralogy of Fallot but others including venticular hypoplasia, TGA and double-outlet right ventricle, have also been reported (7). Other associated anomalies include facial (cleft lip and palate) and skeletal deformities, ventral wall defects and CNS malformations (meningocele and encephalocele).

Thoraco-abdominal ectopia cordis appear to represent a partial form of ectopia cordis and is characterized by partial absence or cleft of the lower sternum, a crescentric midline anterior diaphragmatic defect, a defect of the diaphragmatic parietal pericardium, resulting in a free pericardio-peritoneal communication, an omphalocele like ventral abdominal defect or diastasis recti with partial displacement of the ventricular portion of the heart through the diaphragmatic defect into the epigastrium; and intracardiac congenital heart disease. It has been reported in approximately 37% of the cases of ectopia cordis (5). Toyama reported that at least five cases had no congenital heart disease (6).

Abdominal form of ectopia cordis has been extremely rare and appear to represent a diaphragmatic defect with continued migration of the heart into the abdominal cavity. In some cases, the patients are apparently healthy with no other cardiac disease, and died as adults. In the review of Leca *et al.* 24 cases (11%) were reported to be the abdominal type (5).

In our case report the foetus had thoracic ectopia cordis, omphalocele, anencephaly with a large cystic mass at cranial pole.

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