

Ectopia cordis: a case report

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Ectopia cordis: caso clínico

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Abstract

Introduction: ectopia cordis is a rare congenital malformation, with an estimated incidence of 5 to 8 per million live births. It is defined as a malformation in which the heart is located in an extra-thoracic position. Ectopia cordis may occur as an isolated malformation or associated with other anomalies such as omphalocele, congenital heart disease or integrating Cantrell syndrome. The size and location of the defect influence the prognosis.

Description: we report a case of a 24-year-old nulliparous woman, with no relevant family or personal history, in which the prenatal fetal ultrasound, performed at 21 weeks of gestation, revealed a defect of the anterior chest wall with exteriorization of the heart.

Discussion: fetal echocardiography revealed a severe congenital heart disease. The parents decided to continue the pregnancy, after being duly informed by a multidisciplinary team. Delivery occurred at 37 weeks of gestation but the female newborn died one hour afterwards. Pathological examination confirmed the sonographic findings.

Key words *Ectopia cordis, Heart defects congenital, Prenatal diagnosis, Ultrasonography*

Resumo

Introdução: a ectopia cordis é uma malformação congênita rara, com uma incidência estimada de 5 a 8 por milhão de nados vivos. Define-se como uma malformação em que o coração se localiza numa posição extra-torácica. Pode surgir como malformação isolada ou associada a outras anomalias como onfalocelo, doença cardíaca congênita ou integrando o síndrome de Cantrell. A dimensão e o local do defeito influenciam o prognóstico.

Descrição: descreve-se um caso de uma mulher de 24 anos, nulípara, sem antecedentes pessoais ou familiares relevantes, em que a ultrassonografia obstétrica, realizada às 21 semanas, revelou um defeito da parede torácica anterior com exteriorização do coração.

Discussão: o ecocardiograma fetal revelou uma cardiopatia congênita grave. Os pais decidiram continuar com a gravidez, após de devidamente informados por uma equipe multidisciplinar. O parto ocorreu às 37 semanas, tendo o recém-nascido falecido cerca de 1 hora após o mesmo. O estudo anatomopatológico confirmou os achados ultrassonográficos.

Palavras-chave *Ectopia cordis, Cardiopatias congênicas, Diagnóstico pré-natal, Ultrassonografia*

Introduction

Ectopia cordis is a rare congenital condition that is defined by the abnormal position of the heart outside the thorax associated with defects in the parietal pericardium, diaphragm, sternum, and, in most cases, cardiac malformations. The reported prevalence is 5 to 8 per million live births.¹ The cause of ectopia cordis is currently unknown and most cases are sporadic. Byron classified ectopia cordis into four types: cervical, thoracic, thoracoabdominal and abdominal.² The prognosis is poor and most infants are still born or die within the first few hours or days of life. We present a case report and review the prenatal diagnostic features and management of ectopia cordis.

Description

A 24-year-old nulliparous woman was referred to our maternal and fetal unit when a prenatal fetal ultrasound revealed a cardiac anomaly at the 21st week of gestation (Figure 1). Till then the pregnancy was uneventful. There was no known consanguinity or family history of congenital abnormalities. She had taken no medication during her pregnancy. The

fetal echocardiogram at 23 and 27 weeks of gestation revealed a heart located outside the thoracic cavity, with severe pulmonary valve stenosis, moderate pulmonary artery hypoplasia and a hypoplastic right ventricle. In spite of the poor prognosis being disclosed to the parents by a pediatric cardiologist, they decided not to consent to the recommended amniocentesis but to continue with the pregnancy, owing to religious beliefs. Repeated ultrasound examinations during pregnancy showed a normal growing fetus with a protruding heart. At the 37th week of gestation, a live female infant was born, weighing 3200 g, delivered by caesarean section. Neonatal findings were compatible with *in utero* diagnosis (Figure 2). The neonate died one hour after birth.

The autopsy confirmed the thoracoabdominal wall defect with evisceration of the heart devoid of pericardium and a very small omphalocele. There was also a hypoplastic right ventricle and large left ventricle. The study of the fetal karyotype was normal. The placenta and umbilical cord were unremarkable.

A genetic study of the couple was proposed but they refused.

Figure 1

An ultrasound 2D (a) and 3D (b) image at 21 weeks of gestation showing the fetal heart (arrow) lying completely outside the thorax.



(a)



(b)

Figure 2

Photograph of the neonate demonstrates the midline thoracoabdominal defect.



Discussion

Failure of fusion of the paired cartilage bars of the embryonic sternum leads to a sternal cleft. Its association with a heart located outside the chest wall it is known as ectopia cordis. The heart may be partially or completely outside the thorax. As described by Engum³ and Kaplan *et al.*,⁴ the first report of ectopia cordis was by Haller in 1706. Ectopia cordis was further classified into different types by Weese (1818) and Todd (1836): cervical (3%), cervicothoracic (<1%), thoracic (60%), thora-coabdominal (7%) or abdominal (30%). The thora-coabdominal type is regarded as a distinct syndrome known as Cantrell's pentology, which includes five associated anomalies: distal sternum defect; midline supraumbilical abdominal wall defect; ventral diaphragmatic hernia; defect of the apical pericardium with free communication into the peritoneal cavity; and congenital intracardiac defects.⁵ The various clinical types of ectopia cordis have different prognoses. Cervical and thoracic ectopia cordis are usually fatal within days, because the heart is exposed and malformed. Abdominal ectopia cordis carries a better prognosis, probably because intracardiac abnormalities are rarer and the absence of omphalocele reduces morbidity and mortality.⁶ Ectopia cordis is frequently associated with other congenital defects involving multiple organ systems. Ventricular septal defects and Fallot's tetralogy are the most common associated intracardiac defects, while omphalocele is the most common associated abdominal wall defect. 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.⁷ The prenatal diagnosis of ectopia cordis is carried out using ultrasound, which allows visualization of the heart outside the thoracic cavity. Diagnosis has been reported by Bick *et al.*⁸ and Tongsong *et al.*⁹ at 11 and 9 weeks of gestation, respectively. In our case, prenatal diagnosis occurred at 21 weeks of gestation. The use of three-dimensional ultrasound and its combination with Doppler allows for a more accurate early diagnosis. Magnetic resonance imaging is also becoming commonplace in prenatal evaluation to document and plan for management of complicated congenital anomalies. 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+.⁷ Chromosomal analysis is generally indicated in a patient with prenatally diagnosed ectopia cordis, especially if other anomalies are also identified. Immediate surgical correction of ectopia cordis is often difficult, owing to the inability to enclose the ectopic heart within a hypoplastic thoracic cage. Despite a reported high mortality, numerous successful corrective and palliative cardiovascular operations have

been performed during the neonatal period, as well as during infancy and childhood. Hornberger *et al.*¹⁰ have published data on 13 cases of newborns with ectopia cordis who survived beyond early infancy. In an analysis of 239 cases of ectopia cordis, of which 91 were truly thoracic ectopia cordis, only one survived.³ It is recommended that aggressive surgical procedures should be carried out without delay in the belief that this enhances viability. Overall, the prognosis is poor.

Ectopia cordis is a rare congenital malformation with a poor prognosis. Ultrasonography is of great

value in the prenatal assessment. The ectopia should be precisely located and its classification accurately determined, in view of the different prognoses associated with different types of ectopia cordis. Obstetrical management should include a careful search for associated anomalies, especially cardiac ones, and assessment of fetal karyotype. Pregnancy termination prior to viability and non-aggressive management in the third trimester should be considered and discussed with the parents.

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