

A case report of ectopia cordis and omphalocele

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A rare congenital defect in fusion of the anterior chest wall resulting in an extrathoracic location of the heart. Cantrell's pentalogy is a congenital anomaly resulting from embryologic development defect and consists of the following: A deficiency of the anterior diaphragm, a midline supraumbilical abdominal wall defect, a defect in the diaphragmatic pericardium, congenital intracardiac abnormalities, and a defect of the lower sternum. Here we report a rare case of ectopic cordis with omphalocele.

Key words: Ectopia cordis, omphalocele, scoliosis

Introduction

Ectopia cordis is a rare congenital malformation in which the heart is located partially or totally outside the thoracic cavity. Ectopia cordis may occur as an isolated malformation or it may be associated with body wall defects that affect the thorax, abdomen, or both. With the advances in the medical field and surgical technique, more patients have been successfully treated and have survived. Here, we present a case of ectopia cordis with omphalocele associated with severe kyphoscoliosis.

Case Report

A 24-year-old, unbooked primigravida delivered

spontaneously, a 34-week, male stillborn weighing 1,870 g at Kasturba Hospital, MGIMS, Sevagram. The mother was not suffering from any illness related to pregnancy. There was no history of intake of any teratogens or exposure to unusual environment in antenatal period. The family history was negative for congenital anomalies or genetic abnormalities. There was no history of consanguinity. An anterior thoracoabdominal defect with extrathoracic heart, a cleft sternum, and omphalocele were recognized at birth. The physical examination revealed an exposed heart totally outside of the thoracic cavity without pericardium protection. The abdominal wall defect that caused evisceration of liver, stomach, and the intestines [Figure 1]. Other abnormal features included asymmetrical face with medial epicanthal folds, low set ears, micrognathia, asymmetrical bossing of the skull, and high arched palate. Thorax was small and kyphoscoliosis (thoracolumbar region) of the spine was present. Left sided foot had talipes varus and right foot had valgus deformity. Skeletal survey revealed scoliosis and torsion of the spine. The cranial ultrasound examination was normal. Autopsy confirmed the above skeletal and USG abnormalities. Karyotyping was not done because parents refused to do any genetic testing.

Discussion

Ectopia cordis is a very rare anomaly with an estimated prevalence of 0.079/10,000 births and may occur more frequently in females. It is related to the malformation of the anterior wall of the thorax, with an extrathoracic location of the heart.^[1] In 1958, Cantrell described this syndrome, which occurs sporadically, with variable degrees of expression.^[1] Ectopia cordis

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Figure 1: The anterior view of the thoracoabdominal ectopia cordis with omphalocele

can be classified into five types: 1) Cervical, in which the heart is located in the neck with sternum that is usually intact; 2) thoracocervical, in which the heart is partially in the cervical region, but the upper portion of the sternum is split; 3) thoracic, in which the sternum is completely split or absent, and the heart lies partially or completely outside the thorax; 4) thoracoabdominal, which usually accompanies Cantrell's syndrome; and 5) abdominal, in which the heart passes through a defect in the diaphragm to enter the abdominal cavity.^[2,3] Our case had thoracoabdominal type ectopia cordis in which the bifid sternum, extrathoracic heart, absence of parietal pericardium, and an omphalocele was present.

The formation of the thoracic and abdominal walls is complete in the 9th week of pregnancy and of the heart in the 8th week. Complete or incomplete failure of midline fusion at this embryonic stage can result in a variety of disorders ranging from isolated ectopia cordis to complete ventral evisceration.^[4] The majority of ectopia cordis patients have associated intracardiac defects. Ventricular septal defect, atrial septal defect, tetralogy of fallot, and diverticulum of the ventricle are the most commonly encountered heart lesions.^[5] Ectopia cordis has also been reported with other congenital anomalies such as abdominal wall defects, cranial and facial malformations, cleft lip and palate, anencephaly, hydrocephaly, neural tube defects, pulmonary hypoplasia, genitourinary malformation, gastrointestinal defect, and chromosomal abnormalities.^[6] The defect of the abdominal wall can range from simple diastasis to huge omphaloceles with bowel, liver, and heart. The ectopic

heart may either simply bulge out of the chest or be entirely out of the chest. The diagnosis has been made as early as 17 weeks, but in some cases complicated by oligohydramnios, these cases may be missed entirely.^[7]

The differential diagnosis includes isolated thoracic ectopia cordis, amniotic band syndrome, and body stalk anomalies. The key features for distinguishing these conditions is the position of abdominal wall defect in relation to the umbilical cord insertion, eviscerated organs, the presence or absence of membranes or bands, and associated anomalies. Omphalocele in Cantrell's pentalogy usually involves a midline defect at the umbilical cord insertion. An eccentric large lateral defect and adherence of the placenta to the defect are typically present in body stalk anomalies. The presence of an unexplained ventral wall defect along with extremity deformity with an adherent band suggests amniotic band syndrome.^[8,9]

The prenatal diagnosis is easily made with ultrasound by visualizing the heart outside the thoracic cavity. In view of the poor prognosis, termination of pregnancy can be considered if ultrasound diagnosis is made before viability. The prognosis depends on the degree of the intracardiac involvement and associated malformations, as well as the degree to which the heart is exposed. The majority of neonates die within the first hours of birth. Attempts at surgical correction are already widely performed, with immediate covering of the heart and exposed abdominal contents using silastic prosthesis being recommended. Additionally, a complete evaluation and correction of the intracardiac defects should be performed before closing the abdominal wall.^[10] In conclusion, ectopia cordis with omphalocele is a rare congenital malformation from fatal to nonfatal, therefore it must be adequately evaluated for appropriate prenatal and postnatal management.

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