

Ectopia cordis and amniotic band syndrome

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Abstract

Amniotic band syndrome is a collection of fetal malformations associated with fibrous bands that appear to entangle or entrap various fetal parts in utero leading to deformation, malformation, or disruption. Ectopia cordis is a rare congenital defect in fusion of the anterior chest wall resulting in extrathoracic location of the heart. In this article, we report a rare case of ectopia cordis and amniotic band syndrome in a 26-week-old female fetus. The congenital deformities observed in the present case may be due to earlier rupture of the amnion, ossification defects in bones of the skull, or maternal illness.

Key words: amniotic band syndrome, ectopia cordis, fetus.

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 a larger category of ventral body wall defects that affect the thorax, abdomen or both. Ectopia cordis may occur more frequently in females [1]. The prenatal diagnosis is easily made with ultrasound by visualizing the heart outside the thoracic cavity. Four types of ectopia cordis have been described according to the position of the heart: anterior to the sternum (thoracic – 65%), between the thorax and abdomen (thoracoabdominal – 20%), within the abdomen (abdominal – 10%), or in the neck (cervical – 5%) [2]. The thoracic and thoracoabdominal types are most common, followed by the abdominal and cervical types.

The amniotic band syndrome, proposed to be a sequela of intrauterine rupture of the amnion, results in an unpredictable constellation of fetal anomalies. Deformities of the extremities, thorax, and craniofacial skeleton and soft tissue can occur individually or collectively with varying degrees of severity. The reported incidence of amniotic band syndrome varies from 1: 1,200 to 1: 15,000 live births and the sex ratio appears to be equal. The amniotic band syndrome is found to be sporadic, with no family history and no recurrence in siblings or in children of affected adults [3, 4]. A slight increase in the incidence of amniotic band syndrome in the summer months has been reported. Furthermore, the amniotic band syndrome has been reported in cases in which conception occurred within 1 month of discontinuation of oral contraceptives [3].

The present report is on an unusual case of a 26-week-old female fetus with amniotic band syndrome and ectopia cordis.

Case report

A 26-week-old female fetus was electively aborted due to the presence of multiple anomalies. The CR length of the fetus was 15 cm. The fetus had a small head with a meningoencephalocele. The brain and meninges protruded out of the skull forming two bulges, one anteriorly and one posteriorly (Figure 1). The eyes were rudimentary and the left ear was lobulated. There was a complete cleft palate, associated with a cleft upper lip. The heart and pericardium bulged out of the thoracic wall in the midline. The umbilical cord and the amnion were attached to the entire thoracic wall in the anterior midline and this attachment further extended on the face until the forehead (Figures 1, 2). The lower limit of the umbilical cord attachment was approximately at the epigastric fossa rather than in the lower part of the anterior abdominal wall. The right foot presented a talipes equinovarus condition. The above anomalies would have occurred due to earlier rupture of the amnion, probably within the first 45 days of gestation.

Discussion

The common causes of congenital anomalies are genetic factors, environmental factors or a combination of both. However, for 40 to 60% of congenital anomalies, the aetiology is unknown. The anomalies may be single or multiple and of major or minor clinical significance. According to Moore and Persaud [5], single minor anomalies are present in about 14% of newborns. More than 20% of infant deaths in North America are attributed to birth defects. Of the 3% born with clinically significant congenital anomalies, 0.7% have multiple major defects. Most of these infants die during infancy. Embryos having multiple anomalies are aborted spontaneously by the 6th week of gestation.

Various theories have been proposed for amniotic band syndrome pathogenesis. Streeter [6] in 1930 proposed an endogenous theory in which the primary defect lies in the germinal disc. However, Torpin [7] established that the primary defect could correspond to early rupture of the amnion caused by intrinsic weakness, inflammation, or trauma. This would permit fluid leakage, leading to the introduction of the fetus into the chorionic cavity. The chorion reabsorbs this fluid, stimulating the proliferation of mesenchymal bands. These

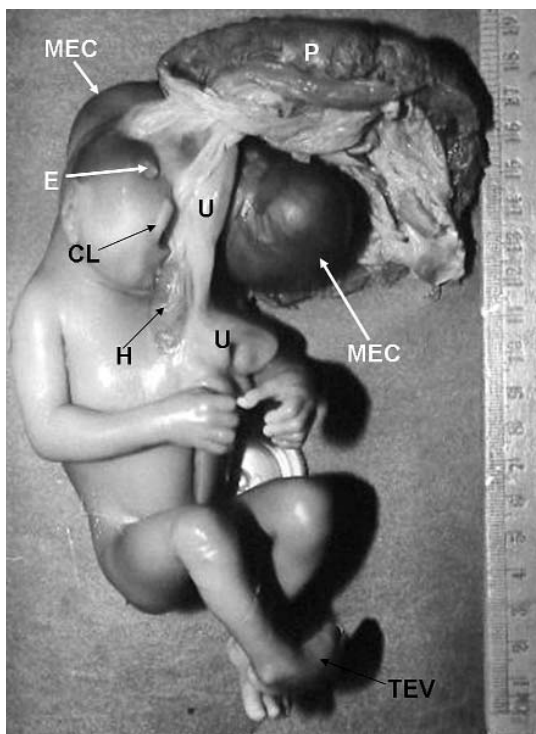


Figure 1. Anterolateral view of the fetus
P – placenta, E – eye, MEC – meningoencephalocele,
U – umbilical cord, H – heart, TEV – talipes
equinovarus, CL – cleft lip

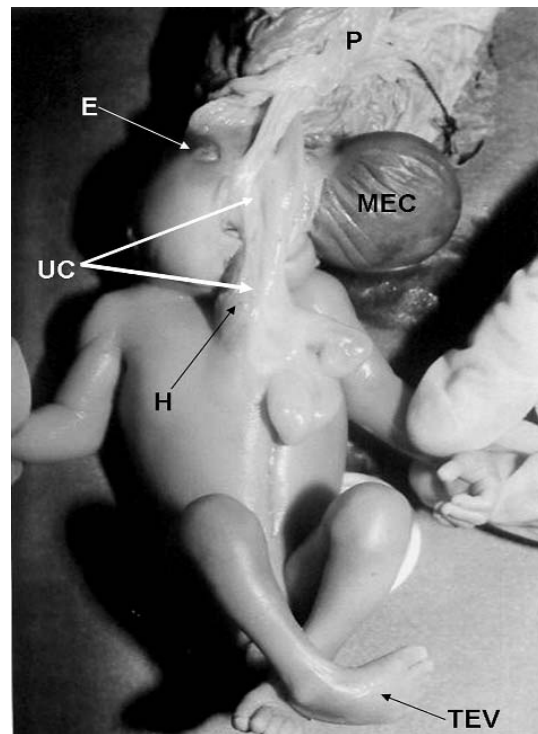


Figure 2. Anterior view of the fetus
P – placenta, E – eye, MEC – meningoencephalocele,
UC – umbilical cord, H – heart, TEV – talipes
equinovarus

entangle the fetus and limit its movements, with subsequent mechanical constrictions.

The incidence of amniotic band syndrome is not high. Familial occurrence is rare. No racial or sex predominance exists. The clinical manifestations are predominantly distal deformities (50%) [8], such as amputation of limbs and digits, syndactyly, pseudosyndactyly, acrosyndactyly (with fenestration), rudimentary toes, and constriction of limbs and digits. Multiple malformations such as clubfoot (50% of patients), leg length discrepancies (39%), other bone abnormalities (12%), special craniofacial defects such as cleft lip and palate, and anencephaly (5%) have been detected in 70% of children with the disorder.

The variability in defect size and severity has been attributed to differences in timing of the amniotic rupture. Early rupture, within the first 45 days of gestation, leads to the most severe defects, including central nervous system and skull defects, facial clefts, cleft lip and palate, limb anomalies, major visceral defects, and abortion. Amniotic rupture occurring at more than 45 days of gestation has fewer central nervous system deformities or clefts and primary defects are usually limited to constriction of isolated limb parts [9]. Routine ultrasonography [10] is important in prenatal diagnosis. Its usefulness in the second half of pregnancy relies on its capacity to distinguish neural tube defects from constrictive bands.

An encephalocele is a region of herniated brain. The term meningocele refers to herniated dura. A combination of herniated brain and dura is called a meningoencephalocele. It is usually caused by ossification defects in bones of the skull (the squamous part of the occipital bone being the most frequently affected bone). Other causes include failure of the neural tube to close (folic acid deficiency), chemical exposures and teratogenic agents [11]. Meningoencephalocele is often associated with anophthalmia or microphthalmia. Meningoencephalocele is common in Southeast Asia. It is of two types, (i) fronto ethmoidal, and (ii) occipital. The case we are presenting had both fronto ethmoidal and occipital bulges of the brain and meninges. It also had rudimentary underdeveloped eyes (Figures 1, 2).

Most cases of cleft lip and cleft palate are multifactorial in origin. Cleft lip occurs in approximately 1:1000 births, more frequently in males (80%) than females. Its incidence is slightly higher with increasing maternal age. The frequency of cleft palate is much lower than cleft lip and occurs in 1:2500 births, more often in females. These are usually caused by drugs, maternal illness, and genetic syndromes.

Ear anomalies are significant from the standpoint of the psychological and emotional trauma. All the frequently occurring chromosomal syndromes and

most of the less common ones have ear anomalies as one of their characteristics.

Ectopia cordis is a rare congenital malformation with a reported incidence of 5.5-7.9/million live births. One of the various causes of ectopia cordis is the abnormal adhesion of the umbilical cord with fetal membranes, which prevents the mesodermal approximation of sternal plates, resulting in pulling of the heart (ectopia cordis). In the current case, the possible reason for ectopia cordis and cleft palate could be the abnormal attachment of the umbilical cord and the amnion along the midline of the thorax and face. Despite significant improvements in neonatal and infant cardiac surgery, operations for ectopia cordis carry an extremely high mortality with only a few reported survivors [12].

Ectopia cordis has been seen more frequently in Turner syndrome and trisomy 18; however, so far there is no evidence that it is a genetically transmissible disease [13].

Talipes equinovarus is observed mainly in males, seen in 1:735 births. It may be due to abnormal position of the legs in the uterus and inheritance (possible oligohydramnios). It is usually present in combination with syndactyly, but in the present case syndactyly was not seen.

The review of the literature suggests that the occurrence of ventral body wall defects and neurological malformations in a single fetus is very rare; there are only two reports in the literature with the above combination [14, 15].

In conclusion, we report a case of amniotic band syndrome and ectopia cordis in a 26-week-old female fetus. The fetus had meningoencephalocele, thoracic type of ectopia cordis, cleft palate, lobulated ear, underdeveloped eyes, club foot and anomalous umbilical cord. The variety of congenital deformities observed in the present case may be due to early rupture of the amnion leading to proliferation of mesenchymal bands and entangling of the fetus, ossification defects in bones of the skull, maternal illness, or psychological and emotional trauma.

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