Ultrasonographic Findings In Pentalogy Of Cantrell With Midline Cleft Lip And Cleft Palate: A Report Of Two Cases

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Citation

K Paritosh, B Alpa, N Ajita, M Suleman. *Ultrasonographic Findings In Pentalogy Of Cantrell With Midline Cleft Lip And Cleft Palate: A Report Of Two Cases*. The Internet Journal of Radiology. 2003 Volume 3 Number 2.

Abstract

We report two cases of Cantrell's Pentalogy detected by ultrasonography at 24 and 25.5 weeks of gestation, respectively and confirmed at autopsy. Cantrell's Pentalogy consists of defects of the lower sternum, anterior diaphragm, midline supraumbilical abdominal wall and diaphragmatic pericardium with ectopia cordis. Our cases showed all these defects along with a bilateral cleft lip and cleft palate in the 24 week fetus. Association of cleft lip and cleft palate with Pentalogy of Cantrell may represent an extension of the basic defect in the migration of mesodermal primordial structures that involve the abdominal and thoracic midline fusion defects of the syndrome to also involve the facial structures, which must therefore be thoroughly looked for in all such cases.

INTRODUCTION

In 1958, Cantrell et al described a syndrome in which a ventral (anterior) diaphragmatic hernia occurred in association with an omphalocele. This syndrome called the Pentalogy of Cantrell 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. Variants of this syndrome have been described. We report two cases of Cantrell's Pentalogy detected by ultrasonography at twenty-four and 25.5 weeks of gestation and confirmed at autopsy, the first case showing an additional feature of a bilateral cleft lip and cleft palate.

CASE REPORT

A twenty-eight year old primigravida presented for routine prenatal ultrasonographic scanning at twenty-four weeks of gestation. The fetus was noted to have a midline umbilical wall defect with a herniated sac containing the liver and bowel (Fig. 1) and the umbilical cord being inserted at the apex of the sac suggestive of an omphalocele, a sternal defect with ectopia cordis, scoliosis and club foot. The head of the fetus was hyperextended and the face was seen touching the anterior wall of the uterus thus making the evaluation of the face difficult. Labor was induced spontaneously and a male gestation weighing 500gms was delivered. Autopsy findings revealed an omphalocele with herniation of the bowel and liver, a midline sternal defect with ectopia cordis; a right-sided club foot, scoliosis and bilateral cleft lip and midline cleft palate. Autopsy features thus confirmed the sonographic features (Fig. 2) except for the cleft palate which was undetected on sonography due to the head of the fetus being hyperextended.

Figure 1

Figure 1: Transverse scan of the fetus depicting herniated thoracic (H = heart) and abdominal (liver and bowel- single arrow) contents.

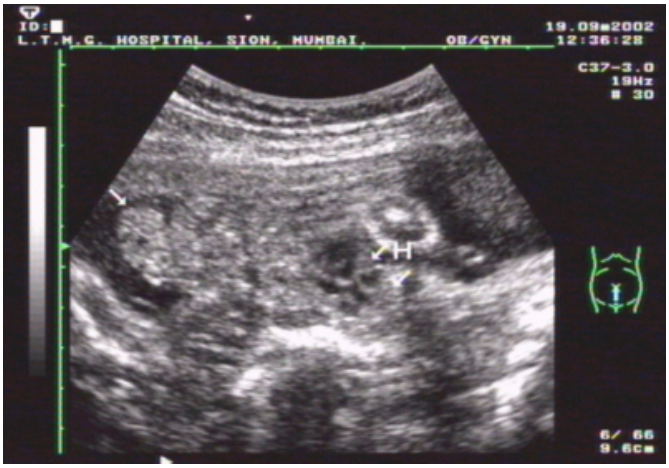
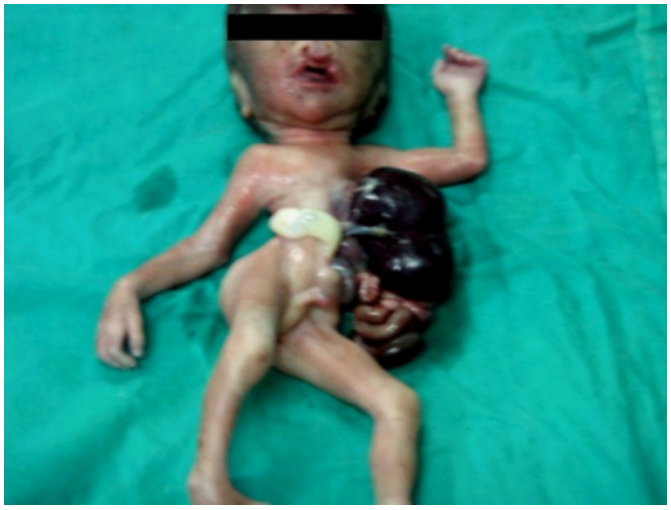


Figure 2

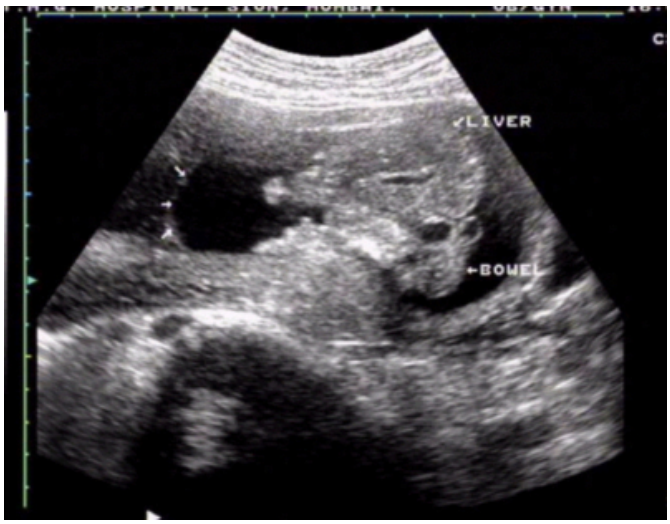
Figure 2: Fetus with omphalocele with herniated liver and bowel, ectopia cordis, club foot, and bilateral cleft lip. The fetus also had a cleft palate.



Another twenty-three year old primigravida also presented for routine prenatal ultrasonographic at 25.5 weeks. This fetus was also live at sonography and had an omphalocele containing herniated bowel and liver (Fig. 3) and a sternal defect with ectopia cordis. A male gestation weighing 700gms was delivered. Autopsy findings revealed an omphalocele with herniation of the bowel and liver and a midline sternal defect with ectopia cordis. Autopsy features thus confirmed the sonographic features in this second case as well. This fetus did not have associated cleft lip or palate and there was no evidence of club-foot.

Figure 3

Figure 3: Antenatal ultrasound of the 25.5-week gestation demonstrating the covering of the omphalocele (arrows on left) and the herniated liver and bowel. Ectopia cordis was also present.



DISCUSSION

In 1958, Cantrell et al[1] described a syndrome in which a ventral (anterior) diaphragmatic hernia occurred in association with an omphalocele. This syndrome called the Pentalogy of Cantrell 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. Few variants of this syndrome have been described[1, 2].

The association of sternal fusion defects with various cardiac, diaphragmatic and anterior body wall defects represents a developmental field complex that includes the Pentalogy of Cantrell and ectopia cordis. Ectopia cordis (extra-thoracic heart) is a rare malformation at the most severe end of a spectrum of anterior body wall defects involving sternal fusion abnormalities[3].

This spectrum of defects is more appropriately categorized as a midline developmental field complex and as such is causally heterogeneous. Evidence for this heterogeneity is suggested by the occasional occurrence of this complex in chromosomal and other multiple malformation syndromes[4].

Variants of the Pentalogy of Cantrell have been described by Toyama. He suggested the following classification for the Pentalogy of Cantrell:

1. diagnosis certain, all five defects present;

2. diagnosis probable, four defects (including intracardiac and ventral abdominal wall) are present; and

3. incomplete, variable combinations of defects present (always including a sternal abnormality)2.

The embryologic defects responsible for the variety of abnormalities present in the Pentalogy of Cantrell are of mesodermal origin. The diaphragmatic and pericardial defects are closely related to either total or partial failure of the transverse septum to develop, whereas the cardiac abnormalities result from faulty development of the epimyocardium. The sternal and abdominal wall defects represent faulty migration of these mesodermal primordial structures. It is thought that these developmental abnormalities occur around day 14 – 18 of embryonic life[5]. The failure of this process is believed to occur due to:

1. Vascular dysplasias resulting in a vascular steal phenomena.

2. Mechanical teratogenesis by amnion rupture, tearing and adhering by the amnion, tissue band adherence causing pressure necrosis and incomplete morphogenesis, mechanical compression secondary to rupture of the chorion or the yolk sac.

3. Genetic mutation, either idiopathic or due to viral infection in the early first trimester or drugs given to the mother6.

In the first of our cases, where there was associated bilateral cleft lip and a midline cleft palate, we believe that this anomaly was due to a migration defect in the primordial mesoderm of the face, which accompanied the ventral body wall defects in the fetus.

Many other associations have been found with Pentalogy of Cantrell, which include amniotic band syndrome with limb anomalies, structural cardiac defects with pericardial effusion, exencephaly, cystic hygroma, infraumbilical defects with cloacal and bladder exstrophy and bilateral inguinal hernias. Concurrent structural and / or chromosomal abnormalities may complicate upto 50 to 75% of cases presenting with omphaloceles and thus it is an indicator for antenatal invasive fetal testing[7, 8]. Omphalocele should be considered pathological only if it persists beyond 14 weeks or its maximum diameter exceeds 1cm in the first trimester of pregnancy[9, 10, 11]. Few cases have been reported in which the pregnancy was continued till term and the fetus delivered by induction of labor. A number of corrective surgeries were done on the neonate for correction of the defects after which a normal growth of the child was achieved by the age of two to three years[5].

CONCLUSION

Thus we conclude that Pentalogy of Cantrell is a spectrum of congenital anomalies, from fatal to non-fatal, which must therefore be adequately evaluated antenatally for appropriate prenatal counseling and postnatal management of the individual cases.

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