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A case of Cantrell syndrome diagnosed in the first trimester

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Summary

Here, the authors report a case of Cantrell's syndrome which was diagnosed by ultrasound at 12th week of pregnancy and confirmed by autopsy. Cantrell syndrome/pentalogy is defined as congenital combination of five main distinctive components: defects at the lower part of the sternum, anterior diaphragm, midline supraumbilical abdominal wall, diaphragmatic pericardium, and ectopia cordis. In the present case, in addition to these anomalies, there was cleft palate and cleft lip at the midline. Association of cleft lip and palate with Cantrell's syndrome may be due to the extension of defective migration of mesodermal primordial structures, which is mainly in abdomen and thorax, towards facial structures. Therefore, in prenatal diagnosis facial anomalies should be examined carefully in all cases with Cantrell syndrome.

Key words: Cantrell syndrome; Congenital defects; First trimester.

Introduction

In 1958, Cantrell *et al.* described a syndrome characterized by the association of ectopia cordis and omphalocele. They called this rare syndrome of midline developmental defects as Cantrell pentalogy, the full spectrum of which consists of an anterior diaphragmatic defect (I), a midline supraumbilical (thoracoabdominal) abdominal wall defect (II), a defect at the diaphragmatic pericardium (III), congenital cardiac anomalies (IV), and a defect of the lower sternum (V) [1]. Many variants of this syndrome has been described in the literature. It is not necessary to diagnose all of the components, and in fact very few cases meet the complete pentalogy spectrum among about 100 case reports of Cantrell syndrome in the literature [2]. In this case report, the authors present a case of incomplete Cantrell syndrome, accompanied by cleft lip and palate anomalies, in which the diagnosis was established by the 12th gestational week ultrasound and confirmed by autopsy.

Case Report

After her first pregnancy ultrasound at the sixth gestational week, a 21-year-old, gravida 2, para 1 woman was admitted for first trimester screening. In prenatal ultrasonography, ectopia cordis in which the fetal heart was seen as protruding from the defect at the anterior chest wall and fetal midline facial defect was observed. As these findings supported the diagnosis of Cantrell syndrome, the prognosis of pregnancy was discussed with the parents. Patient was referred to a tertiary center. As family requested termination of pregnancy because of the poor prognosis, induction was began with vaginal administration of misoprostol 200 mcg every four hours. Six hours after the first dose of misoprostol, patient underwent a complete abortion. On postabortive gross examination, extensive anterior chest wall defect, partial ectopia cordis, cleft palate, cleft lip, and nasal dysplasia were observed. The patient was discharged the morning after abortion without complications. Autopsy examination revealed an anterior diaphragmatic defect, a diaphragmatic pericardial defect, lower sternum defects, ectopia cordis including large ventricular septal defect, cleft palate, cleft lip, and nasal dysplasia. There was no evidence of a supraumbilical abdominal wall defect.

Discussion

Cantrell pentalogy is a rare syndrome of congenital midline developmental failure, the full spectrum of which consists an anterior diaphragmatic defect, a midline supraumbilical abdominal wall defect, a diaphragmatic pericardial defect, congenital cardiac anomalies, and a defect of the lower sternum. The incidence was reported from 1: 65,000 to 1: 200,000 [3]. Defined as the displacement of heart outside the thorax, ectopia cordis is an anomaly usually fatal in the first few days after birth. Complete ectopia cordis is defined if the heart has no pericardial cover outside the thorax while, partial ectopia cordis is diagnosed if pericardium and skin covers the heart outside the thorax [4]. There is accompanying ventricular septal defect (VSD) in all cases of ectopia cordis, while atrial septal defect (ASD) is present in 53% [5].

The pathogenesis of Cantrell syndrome is not fully understood. Cantrell *et al.* proposed an embryologic developmental failure of a segment of the lateral mesoderm

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around 14-18th days of gestation [1]. As a result, the development of transverse septum of the diaphragm fails and a pair of mesodermal folds located in the upper abdomen do not migrate to the ventromedial. Organs eviscerate through sternal and abdominal wall defects.

The most common abdominal wall defect in the Cantrell syndrome is omphalocele, which was absent in the present case [6]. Anterior diaphragmatic and pericardial defects are also common. The most common cardiac anomaly reported is thoracoabdominal ectopia cordis [5]. However, ectopia cordis is not a main component of pentalogy of Cantrell, the syndrome can be diagnosed without ectopia cordis [5]. In the present case, ectopia cordis was present in association with a large VSD. Other anomalies that can accompany the five main components of the syndrome have been reported as: craniofacial malformations (cleft lip and palate, anophthalmia, microphthalmia, hypertelorism), cystic hygroma, meningocele, anencephaly, pulmonary hypoplasia, vertebral anomalies, finger anomalies, pes equinovarus, adrenal agenesis, renal agenesis, intestinal malrotation, and hypospadias [1-8].

Diagnosis of Cantrell syndrome can usually be made by prenatal ultrasonography in the first trimester [9]. While presence of ectopia cordis and omphalocele facilitates early prenatal diagnosis, paucity of the defect may delay the diagnosis to the second trimester [7]. Pentalogy of Cantrell should be ruled out in a fetus with omphalocele. If pericardial effusion is seen, anterior diaphragmatic hernia and diaphragmatic pericardial defects should be suspected, and in the presence of them, a detailed ultrasound should be performed for the aforementioned findings of pentalogy of Cantrell [8]. Prenatal magnetic resonance imaging, which is used more often in the second trimester, may facilitate the diagnosis of fetal anomalies [10]. Because of the infrequency and variations in the clinical spectrum, both the diagnosis and gaining experience of the surgeons become more difficult.

Cantrell pentalogy has been described in many varieties in the literature since components of this syndrome can occur in different clinical severity and combinations. It is not essential to diagnose all of the findings, and very few cases meet the original pentalogy spectrum among about 100 case reports described in the literature so. In the present case, supraumbilical midline abdominal wall defect was not observed. In addition to the other four anomalies of the spectrum, midline cleft palate and cleft lip, and nasal dysplasia was present. Association of cleft lip, cleft palate, and nasal anomalies with Cantrell's syndrome may be due to the extension of defective migration of mesodermal primordial structures, which mainly leads to midline fusion defects in abdomen and thorax, towards facial structures. Consequently, in the present authors' opinion, facial anomalies should be carefully examined in all cases with Cantrell syndrome.

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