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Case report-Olgu sunumu

Pentalogy of Cantrell

Cantrell pentalojisi

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Abstract

Ectopia cordis is a rare malformation presenting as an isolated lesion or as part of the Cantrell's pentalogy syndrome characterized by midline closure defects. A preterm male baby who did not have antenatal follow-up was born by caesarean section with evisceration of the heart and major parts of the intraabdominal organs. Combination of these findings suggested the diagnosis of Cantrell's pentalogy. Despite intensive care, the patient died soon after the birth due to the poor clinical state. In order to make contribution to literature about this subject, this rare case of neonate with Cantrell's pentalogy was reported. Antenatal diagnosis, appropriate antenatal care and delivery in a hospital that is capable for treatment is important for a better prognosis.

Keywords: Abdominal wall defect, Cantrell's pentalogy, ectopia cordis, newborn

Özet

Ektopia kordis, izole bir lezyon olarak ya da orta hat kapanma defektleri ile karakterize Cantrell pentalojisinin bir parçası olarak ortaya çıkan nadir bir malformasyondur. Antenatal takibi olmayan preterm bir erkek bebek, kalp ve karın içi organların büyük bir kısmı vücudun dışında bir halde sezaryen ile doğurtuldu. Hastadaki bu bulguların birlikteliği Cantrell pentalojisi tanısını düşündürdü. Yoğun bakım desteğine rağmen klinik durumu kötüleşen hasta doğumundan kısa bir süre sonra kaybedildi. Cantrell pentalojisi tanısı konulan bu nadir yenidoğan vakası konu hakkında literatüre katkı sağlamak amacıyla sunuldu. Antenatal tanı, uygun antenatal bakım ve doğumun tedavi olanakları olan donanımlı bir merkezde yapılması ile prognoz daha iyi olacaktır.

Anahtar sözcükler: Karın duvarı defekti, Cantrell pentalojisi, ektopia kordis, yenidoğan

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Introduction

Ectopia cordis (EC) is defined as an anomaly in which the fetal heart lies outside the thoracic cavity. This rare condition is usually associated with Cantrell's pentalogy. The pentalogy of Cantrell was first described in 1958 [1]. The full spectrum consists of five anomalies: a deficiency of the anterior diaphragm, a midline supraumbilical abdominal wall defect, a defect in the diaphragmatic pericardium, various congenital intracardiac abnormalities, and a defect of the lower sternum [1-3]. It has an estimated incidence of 5.5 per 1 million live births [4].

Because of its rarity, we report a case of neonate with Cantrell's pentalogy and present a brief review of the literature.

Case report

A male baby was born at 31 weeks' gestation by emergent caesarean section due to fetal distress, from a 19-year-old gravida 1, para 1 mother, with evisceration of the heart, liver, spleen, stomach and a major part of the intestines. There was no history of trauma or radiation exposure, or infection on prenatal history. The mother has never been examined by ultrasonography (US) during pregnancy. However, on admission, ultrasonographic examination of mother revealed evisceration of the heart and intraabdominal organs in the fetus, but no other additional anomaly. Unfortunately, the emergent clinical state of fetus did not permit to perform an echocardiographic examination. On family history, parents were first degree cousins, and there was not any history of congenital abnormality in the family. The patient's birth weight was 1245 g, and his Apgar scores were 3 and 5 at one and five minutes, respectively. After resuscitated and intubated in the operating room, he was transferred to our neonatal intensive care unit (NICU). On admission, the patient's general condition was very poor with cyanosis of tongue, lips, and skin. Oxygen saturation levels were between 50% and 60% with a body temperature of 35°C, and pulse rate 104 beats/min, his arterial blood pressure was 45/18 mmHg (mean 23 mmHg). On physical examination, his heart was lying outside the thoracic cavity and there was a supraumbilical anterior abdominal wall defect in which liver, spleen, stomach and a major part of the intestines were eviscerated from there (Figure 1). Other than this, no external deformity was found in any other part of the body. Arterial blood gas analysis showed a pH of 7.27, PCO2 53.8 mmHg, PO2 42.9 mmHg, HCO3 18.4 mmol/L, and BE -7.6. Chromosome analysis showed a normal male karyotype, 46XY.

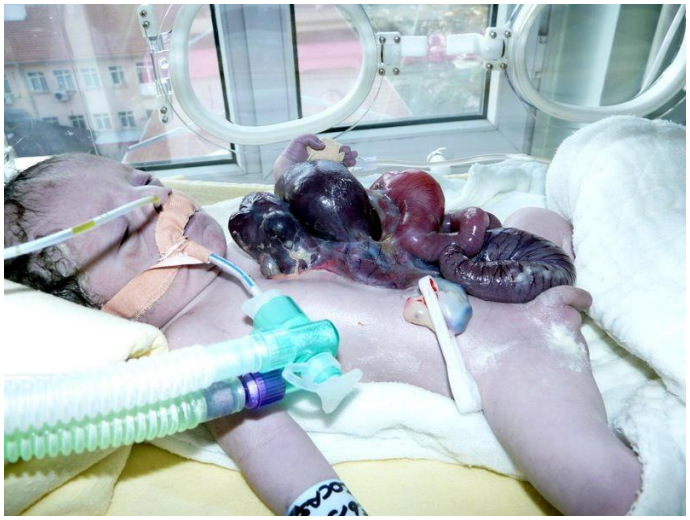

Figure 1. Note that the heart lying outside the thoracic cavity with evisceration of the liver, stomach and a major part of the gastro-intestinal tract

Co-existence of EC and abdominal wall defect suggested us the diagnosis of Cantrell's pentalogy. After admission to NICU (Newborn Intensive Care Unit), he received mechanical ventilation, appropriate amount of parenteral fluid and broad spectrum antibiotics. Appropriate measures were taken to correct the hypothermia and avoid dehydration. Since he was hypotensive, inotropic support was given with dopamine and dobutamine. The patient's poor clinical state, infection risk, and early death of the present proband impeded a complete evaluation of the cardiac status. While it was planned to transport the patient to another hospital capable for emergent operation, the surfaces of the eviscerated organs were covered with sterile warm wet gas compresses. These compresses were changed with another to avoid hypothermia and dehydration. Despite inotropic support, the child remained to be hypotensive with oxygen saturation levels maintained between 50 and 60%. He died on the 4th hour of his life. Unfortunately, the parents did not give the permission for necropsy.

Discussion

Pathogenesis of Cantrell's pentalogy has not been fully elucidated yet. Cantrell et al. [1] have suggested an embryologic developmental failure of a segment of the lateral mesoderm around the gestational age of 14-18 days. Therefore, the transverse septum of the diaphragm does not develop, and the paired mesodermal folds of the upper abdomen do not migrate ventromedially. Hence, organs eviscerate through in sternal and abdominal wall defects. Depending on the location of the protruding heart and on the extent of the body wall defect, EC may be classified into cervical, thoracic, thoracoabdominal, or abdominal types [3, 5]. As seen in our case, its' thoracic or thoracoabdominal type are associated with Cantrell's pentalogy [6].

Most cases of the EC appear as isolated, sporadic defects while others are associated with chromosomal abnormalities (trisomy 18). Various associated anomalies have been reported that are craniofacial and central nervous system anomalies such as cleft lip and/or palate, encephalocele, hydrocephalus, and craniorachischisis; limb defects such as clubfoot, absence of tibia or radius, and hypodactyly; and abdominal organ defects such as gallbladder agenesis and polysplenia [4, 7-9]. In our case, normal 46XY karyotype has been shown and, based on prenatal US and physical examination, no any additional anomaly other than that of Cantrell's pentalogy has been determined.

Cantrell et al. [1] reported that intracardiac anomalies seen in the pentalogy of Cantrell are that of ventricular septal defect (100%), atrial septal defect (53%), tetralogy of Fallot (20%), and ventricular diverticulum (20%) [1]. Although an echocardiographic evaluation could not be performed in our case, sustained hypotension resistant to inotropic treatment and constant hypoxemia without hypercarbia in spite of respiratory support suggested the presence of possible additional complex intracardiac defects.

Many variants of Cantrell's pentalogy have been described according to the postulated embryological developmental defects. These various types could be classified as follows: Class 1, also called as exact diagnosis, in which the five defects are present; Class 2, also called as probable diagnosis, in which the four defects (including intracardiac and abdominal wall defects) are present; and Class 3, incomplete diagnosis, this is the combination in the defects (usually accompanied by sternal defects) [1, 2, 10, 11]. Although a complete cardiac evaluation could not be performed and small defects of the diaphragm and pericardium are extremely difficult to diagnose accurately [5], very poor clinical course seen in our case suggested us that he possibly belongs to class 1 embryological classification. With prenatal US, the pentalogy of Cantrell usually can be diagnosed in the first trimester of pregnancy [12]. In a fetus with omphalocele, pentalogy of Cantrell should be ruled out [13]. However, in our case, diagnosis could be made shortly before delivery because there was not any antenatal follow-up. Nevertheless, a detailed sonographic evaluation intended for detecting additional anomalies and intracardiac defects could not be performed because of emergent state. After birth, echocardiography is essential for diagnosis of associated cardiac anomalies. Other features of the pentalogy of Cantrell and known associated anomalies can be diagnosed by conventional radiography or by US [5]. However, in our case, very poor clinical picture and early death did not allow us for comprehensive evaluation of the possible additional anomalies. Isolated thoracic type EC or EC associated with amniotic band syndrome are the conditions that should be considered in the differential diagnosis [14]. Amniotic band syndrome was excluded in our case based on the lack of any destructing lesions those are typical for amniotic band syndrome. The treatment of the pentalogy of Cantrell consists of corrective or palliative cardiovascular surgery, correction of ventral hernia and diaphragmatic defects and correction of associated anomalies. The best treatment strategy depends on the size of the abdominal wall defect, the associated heart anomalies, and the type of EC. As seen in our case, the prognosis is poorer and the mortality rate is very high in the patients with complete form of the disease, and in patients who have associated anomalies [5]. Therefore, before delivery, the patients with

complex anomalies should be referred to a hospital that is capable for treatment. A prenatal medical team consists of a gynecologist, a neonatologist, a pediatric cardiologist, a geneticist, and a pediatric surgeon who should use their expertise for choosing the best approach to this severe disorder [5]. But, if antenatal diagnosis is lacking, as seen in present case, qualified medical assistance could not be given to the neonates. Therefore, antenatal diagnosis and an appropriate antenatal care are very important for prevention and management of the problems both in mother and baby.

In conclusion, co-existence of EC and anterior abdominal wall defects should consider the diagnosis of the Cantrell's pentalogy. This lethal syndrome requires prompt medical and surgical interventions soon after birth. Therefore, if a case of Cantrell's pentalogy is antenatally diagnosed, referral to a hospital that is capable of multidisciplinary approach should be planned before birth.

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