Ankara Üniversitesi Tıp Fakültesi Mecmuası 2007, 60(3)

A Severe Form of Cantrell` s Pentalogy with Complete Ectopia Cordis

Komplet Ektopiya Kordisli Cantrell Pentalojisi Olgusu

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Cantrell's Pentalogy (CP) is a rare congenital syndrome characterized by defects of lower sternum, anterior diaphragm and pericardium with complete or partial ectopia cordis, an omphalocele and congenital intracardiac defects. Few cases manifesting all the defects have been reported, however, only a small number have survived as the outcome mainly depends on the cardiac malformation. Herein, we report a case of most severe form of CP manifesting all defects and associated with complete thoracoabdominal ectopia cordis to discuss the challenges in management of this uncommon entity.

Key Words: ***Ectopia cordis, Cantrell`s pentalogy, omphalocele***

Cantrell pentalojisi, komplet veya parsiyel ektopia kordis ile birlikte alt sternum, anterior diyafragma ve perikard defektleri, omfalosel ve konjenital intrakardiyak defektlerle karakterize olan ve seyrek görülen bir konjenital sendromdur. Tüm bu defektlerin birlikte görüldüğü çok az sayıda vaka rapor edilmiştir. Bununla birlikte sonuçlar ana olarak kardiyak malformasyona bağlı olduğundan çok az sayıda hasta sağ kalabilmiştir. Burada, sözügeçen tüm defektlerin ve eşlik eden komplet torakoabdominal ektopia kordisin tespit edildiği Cantrell pentolojisinin en ağır formu ve bu nadir görülen durumun tedavisindeki fikir ayrılıkları rapor edilmektedir.

Anahtar Kelimeler: ***Ektopia kordis, Cantrell pentalojisi, omfalosel***

Ectopia cordis is a rare congenital malformation occurring in 5.9-7.9 million live births and is characterized by complete or partial displacement of the heart out of thoracic cavity(1-6). Cantrell and colleagues described the association of complete or partial ectopia cordis with defects of lower sternum, anterior diaphragm and parietal pericardium, midline supraumbilical abdominal wall defects and congenital intracardiac defects in 1958, since called, Cantrell's Pentalogy (CP) (7). Few cases manifesting all the defects have been reported (4,5,7,8,9), however, only a small number have survived as the outcome mainly depends on the cardiac malformation and operations for ectopia cordis still carry extremely high mortality despite the improvements in neonatal surgery (8). Herein, we report a neonate manifesting all defects of CP and complete ectopia cordis to discuss the challenges in management of this uncommon entity.

Case Report

A 6 hour-old, 2000g, female infant delivered vaginally at 38 weeks gestation to a G2 P2 mother was referred from a maternity hospital because of having extrathoracic heart. Prenatal investigations were not present. Apgar scores were 5 and 8 at 1 and 5 minutes. The patient was moderately cyanotic newborn female with complete

Received: 08.03.2007 • *Accepted:* 03.05.2007

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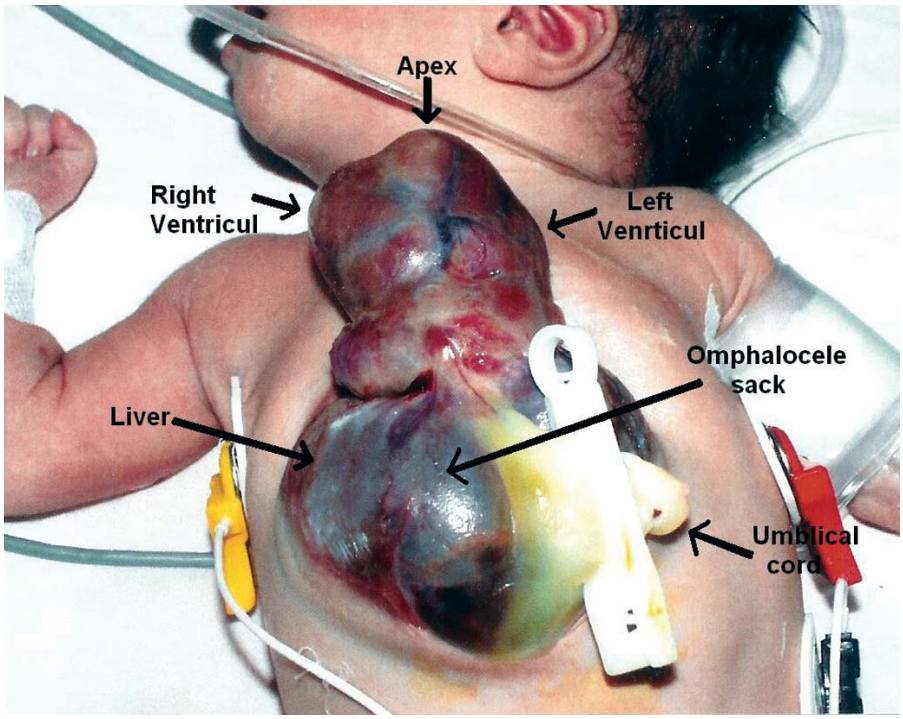

Figure 1. Photograph of the patient with CP and ectopia cordis obtained at initial presentation

thoracoabdominal ectopia cordis and inverted positioning of the heart. The heart rate was 194 beats/min, systolic/diastolic blood pressure was 82/55 mmHg and respiratory rate was 65 breaths/min. There was a paradoxic retraction of the anterior chest wall with each inspiratory effort. Pulse oximetry showed oxygenated hemoglobin saturation of 85 %. There was a midline defect in the anterior chest wall extending from suprasternal notch superiorly to the umbilicus inferiorly. The heart was visibly pulsating out side the thoracic cavity without any skin or pericardial cover. The apex of the heart and ventricles were positioned superiorly, and the atriums were lying inferiorly. Four pulmonary veins, inferior and superior vena cava, and aortic arch were visible and arcus aorta was lying inferiorly. Additionally there was an eight cm diametered omphalocele in which a significant part of liver was observed (Fig 1).

Initial laboratory evaluations including hemoglobin level, white blood cell count, liver and kidney function tests were with in normal limits. Blood gas analysis revealed a pH of 7.15, PO_2: 48 mmHg and PCO_2: 62 mmHg with base excess of –9.1mmol/L. The patient was promptly intubated and ventilated. Further work-up included an echocardiogram demonstrating large atrial and ventricular septal defects and pulmonary stenosis. The patient was consultated with the department of Pediatric Cardiovascular Surgery. Corrective heart operation and repositioning the heart in to the thoracic cavity was not considered during initial surgery. Primary surgical repair was decided to provide soft tissue coverage to abdomen and heart.

After dissection of the myocardium and omphalocele sac from the skin edges were carried out, the anterior diaphragmatic defect was noted. The right and left hemidiaphragm were lying separately and herniation of the liver into the thoracic cavity from the diaphragmatic defect was present. After reduction of the liver into the abdominal cavity, the right and the left hemi diaphragms were sutured at the midline. Coverage of the anterior chest wall was tried by careful dissections of the myocardium and the skin edges, followed by elevation of full thickness skin flaps. The edges were approximated with close monitoring of hemodynamics, necessitating a Gore-Tex (W.L. Gore & Assoc, Naperville, IL) pericardial substitute extension. The abdominal defect was also repaired using Gore-Tex (W.L.Gore & Assoc, Naperville, IL) fascial substitute for facial defect reconstruction with elevation of intraabdominal pressure 10 cm H_20. After a successful hemodynamic palliation and respiratory support during the first 24 hours, the baby suddenly developed hemodynamic that led to death.

Autopsy confirmed the clinical diagnosis of CP with the following intracardiac defects: Large ventricular septal defect, atrial septal defect, right ventricular double outlet and severe narrowing of infundibulum. There was no hypoplasia of the lungs.

Discussion

The pathogenesis of CP has not been understood since its first description. Cantrel et al. postulated these malformations to result from developmental failure in differentiation of a segment of intraembryonic mesoderm between splanchnic and parietal planes around 14-18 days of embryonic life (7). It has been proposed that compression

of the thoracic cavity, resulting from rupture of the chorion /yolk sac around 21 days of gestation prevents proper midline fusion of the developing chest wall (1,3-5,9-11). That may result in delayed retraction of the bowel in embryo with defective formation of ventral body wall and defective formation of septum transversum (1,3-5, 9-11).

Ectopia cordis is characterized by complete or partial displacement of the heart out of thoracic cavity (1-6). It is classified as either cervical, thoracic, thoracoabdominal or abdominal type, relative to the position of the displacement of the heart (1-6). The thoracoabdominal is the most common form and is commonly associated with CP (1-6). The overall survival of the thoracoabdominal form is reported as 50% depending on other associated congenital anomalies and the type of heart defect (1-6). The thoracic type is usually associated with dismal prognosis(1-6). Common cardiac congenital anomalies associated with ectopia cordis are ventricular septal defect (60-100%), atrial septal defect (53%), left ventricular diverticulum (9-20%), and tetralogy of Fallot (9.3 -20%) (1-6). Rarely ectopia cordis can occur with structurally normal heart.

Although there is controversy as to the treatment of CP, surgical strategy mostly depends on the type of ectopia cordis, associated heart anomalies and the size and content of the omphalic defect (1,4,5,8) In milder cases of thoracoabdominal ectopia cordis and CP, in which the omphalocele is small or nonexistent or heart is protruding through an anterior diaphragmatic defect and is covered by skin and soft tissues, corrective heart operation, ventral hernial and the diaphragmatic defect repair can be performed at the same time (1-5,8,12,13). In severe forms of CP in which a large omphalocele, herniation of the liver into the chest, pulmonary hypoplasia and complete ectopia cordis are present, a two-stage repair is advocated (1-5,8). The goal of the first operation in the neonatal period is to provide urgent soft tissue coverage to abdomen and heart and closure of the pericardiopleural communication.

In majority of the successful cases coverage of anterior chest wall with skin flaps or use of prosthetic patches were well tolerated (1,13,14). During the second stage corrective heart operation is performed between 6 months and 2 years of age. Intracardiac repair with concomitant chest wall reconstruction by means of a neosternum formed by ribs and perichondrium and reduction of the heart into the thoracic cavity could be performed (14-15).

Despite modern surgical standards, severe form of CP and complete ectopia cordis represents a challenge to the surgeon because of the wide range of the anomalies and severity of the abdominal and cardiac malformations. Any significant extracardiac defects, pulmonary hypoplasia, large abdominal defects, cerebral defects and herniation of liver and bowel into the thoracic cavity worsen the prognosis of CP. Although there are some reports of successful cases of complete ectopia cordis, survival after birth averages 36 hours; intracardiac defects are associated in 80% of the cases and all unoperated patients died (1,2,3,10). The present case represents the one of most severe form of CP and complete ectopia cordis with inverted positioning of the heart and severe intracardiac defects consisting of double outlet of right ventriculi with atrial and ventricular septal defects and narrowing of the infundibulum. Urgent soft tissue coverage of the heart and abdomen was carried out to prevent hypothermia, fluid loss, cardiac desiccation and trauma. However, coverage of the chest wall led to hemodynamic instability and compromise of cardiac function with fatal outcome although meticulous hemodynamic support with positive inotropic agents and mechanical ventilation was tried. Hemodynamic palliation is utmost important during soft tissue coverage of the heart. The other crucial problem seems to be the avoidance of high intraabdominal and intrathoracic pressures postoperatively as they cannot be tolerated in the presence of severe cardiac malformation. Coverage of the ectopic heart should be tried with close hemodynamic monitoring, and if there is any decompensation when the edges of full thickness skin flaps are approximated, a Gore-tex extension should be attached and reduced slowly over ensuing weeks. In attempting to reposition the heart into mediastinum angulation of the great vessels and fatal compromise of the cardiac function may easily occur especially if apex of the heart is directed cephalad. Angulation of great vessels can be minimized if they are dissected as free as possible. Intracardiac defects had major influence fatal outcome of the present case. Some surgeons consider it preferable to attempt cardiac repair prior to repair of the abdominal and chest wall defects believing that infants with congenital heart lesions do not tolerate abdominal and thoracic wall repair unless cardiac defect is

repaired (13).

Depending on our experience, the major challenge in management of severe cases of CP with complete ectopia cordis is the difficulty to maintain hemodynamic stability because of the major intracardiac defects. However, despite poor outcomes, surgical repair should still be attempted on these cases because the best and only chance

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