CASE REPORT

Successful Surgical Repair of Pentalogy of Cantrell at 14 Months of Age

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ABSTRACT

Pentalogy of Cantrell with ectopia cordis is a rare congenital anomaly, first described in 1958 by Cantrell, has a reported incidence of around 5 - 10 cases per one million live births with wide variety of clinical presentations. We are reporting a child with ectopia cordis along with cleft lower sternum, upper abdominal wall defect, ectopic umbilicus and diaphragmatic defect. Echocardiography in first month of life revealed a restrictive perimembranous ventricular septal defect and a small patent Foramen Ovale, both closed spontaneously in infancy. CT angiography at 10 months of age revealed a defect in the thoracic and abdominal walls along with herniation of left ventricular apex into epigastrium. The two ventriculi formed a tail that looked like a crocodile. This patient underwent surgical correction at our institution at 14 months of age and recovered well with no residual issue.

Key Words: *Pentalogy of Cantrell. Ectopia cordis. Abdominal wall defect.*

INTRODUCTION

Pentalogy of Cantrell or thoraco-abdominal ectopia cordis is a rare congenital syndrome characterized by five abnormalities including abdominal wall defect (usually omphalocele), lower sternal defect, diaphragmatic pericardial defect, anterior diaphragmatic defect and intracardiac abnormalities.[1-5] First described by Cantrell in 1958, the syndrome occurs sporadically, with variable degrees of expression[6] and is reported from developed countries in detail. Common cardiac defects associated with Pentalogy of Cantrell include ventricular septal defect, tetralogy of Fallot and atrial septal defect.[1] Surgical repair remains challenging as it involves placement of the heart within the thoracic cage and complete repair of the diaphragmatic and abdominal defect as well as associated intra-cardiac defects.

We are reporting a case of Pentalogy of Cantrell who was successfully operated upon at our institution, at 14 months of age with uneventful recovery.

CASE REPORT

A child first presented at 20 days of age with a pulsating mass in the supra-umbilical region. The baby was born by spontaneous vaginal delivery in another hospital at term and was acyanotic with no respiratory distress. Antenatal history was unremarkable and there was no maternal history of gestational diabetes, hypertension, smoking, TORCH infections or history of maternal drug exposure during pregnancy. Local examination showed a spherical, pulsating mass 10 x 10 cm comprising of a lax but intact pigmented skin overlying the heart that was herniating out in the supra-umbilical region through a defect in the anterior diaphragm (Figure 1a). The anterior chest wall was intact and there was no scoliosis or kyphosis.

Chest X-ray showed a rather narrow midline cardiac Silhouette with no evidence of lung hypoplasia. ECG showed RsR' pattern in the leads II, III and aVF. Two dimensional echo performed on the first visit demonstrated a small perimembranous ventricular septal defect (VSD) that was restrictive in nature with a small persistent foramen ovale (PFO) with otherwise normal heart. Both ventricles had their apices herniating from the diaphragm (diverticuli) in cross-sectional and long-axis two dimensional views taken at the diaphragm (Figure 1b). Contrast enhanced CT pulmonary angiography at 10 months of age revealed a ventral

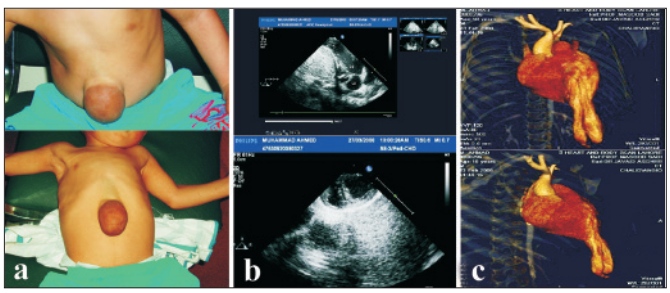

Figure 1 (a,b,c): (a) Patient picture with Pentalogy of Cantrell **(b)** 2D-Echocardiography showing both diverticuli in cross sectional and long axis **(c)** CT Angio showing defect in the lower thoracic and upper abdominal wall continuous with a defect in the anterior diaphragm and out pouching of left ventricular apex herniating into epigastrium.

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defect in the lower thoracic and upper abdominal wall continuous with a defect in the anterior diaphragm. A 45 x 20 mm outpouching of left ventricular apex herniating into epigastrium along with umbilical hernia was also evident. The two ventriculi formed a tail that looked like a crocodile (Figure 1c).

He remained on regular follow-up for the next one year. VSD and PFO spontaneously closed before first birthday. The patient was admitted at 14 months of age at our institute for surgical repair which included resection of the diverticulum and placement of the heart within the thoracic cage and complete repair of the diaphragmatic and abdominal defect. This case was operated by Professor William Novick in July 2008 at AFIC / NIHD, Rawalpindi, Pakistan. The left ventricle was detached from the pericardium and hernial sac. Left pleura was opened and a pericardial pouch was created with pericardial membrane. Ventricular extension was placed in this pouch. Umbilical hernia was repaired. He made uneventful recovery and was discharged on the 10th postoperative day. He is doing well and last follow up was made 2 years after surgery.

DISCUSSION

Pentalogy of Cantrell was first described by Cantrell *et al*. in 1958.[1,6] Cantrell's pentalogy with ectopia cordis is a rare and lethal congenital anomaly, with reported prevalence of 5 - 10/1 million live born infants.[1,5,7] The pentalogy of Cantrell includes a midline supra-umbilical abdominal defect, a defect of the lower sternum, a deficiency of the diaphragmatic pericardium, the anterior diaphragm and congenital cardiac anomalies.[1,6,7] Intracardiac defects includes ventricular septal defect (100%), tetralogy of Fallot, atrial septal defect, pulmonary stenosis or atresia, transposition of great arteries, tricuspid atresia, truncus arteriosus, atrio-ventricular septal defects, anomalous pulmonary venous return and right ventricular diverticulum.[3,7,8] Left ventricular diverticulum is present in 20 - 50% of the cases.[8] This case had both the left and right ventricle diverticulum herniating through the supra-umbilical abdominal defect (Figures 1a and 1c). Furthermore, the patient had a restrictive perimembranous VSD that spontaneously closed by one year of age leaving him to have an otherwise functionally normal heart. Various other associated anomalies including craniofacial and central nervous system anomalies are reported,[4] but in this case no such abnormalities were present.

The exact aetiology is still not clear with some reports of familial forms.[2] The sternum, abdominal wall, pericardium, and part of the diaphragm arise from somatic mesoderm, while the myocardium arises from splanchnic mesoderm. An event occurring prior to differentiation of the mesoderm into these two layers could produce defects in all of the involved structures, as seen in pentalogy of Cantrell. Although a specific aetiology is unknown, the timing of the event or insult is thought to be between 14 and 18 days after conception.[5] One of the proposed embryogenesis process postulates that the failure of the lateral mesodermal folds to migrate to the midline, causing the sternal and abdominal defects, and failure of the septum transversum to develop, result in defects in the anterior diaphragm and pericardium.[6] Long-term prognosis is generally poor in complete form and is not dependent upon intracardiac defects.[4] In view of the poor prognosis, termination of pregnancy can be considered if ultrasound diagnosis is made before viability. In patients choosing to continue the pregnancy, there is no data indicating improved or changed outcome with caesarean delivery.[9] After delivery, surgical repair is the best treatment option and greatly depends upon defect size and associated cardiac anomalies.[4]

Surgical management includes corrective or palliative cardiovascular surgery, correction of ventral hernia and diaphragmatic defects and correction of associated anomalies.[3,4] Repair of the sternal, diaphragmatic, and pericardial defects can be attempted at the same time. Surgical correction is often difficult secondary to hypoplasia of the thoracic cage and inability to enclose the ectopic heart. Some affected infants have respiratory insufficiency secondary to pulmonary hypoplasia. Resection of the ventricular diverticulum is indicated early in life because of the risk of spontaneous or traumatic rupture and sudden death by tachyarrhythmias (6 - 20%).[10] Severe abdominal wall anomaly with a relatively stable cardiac defect may need correction of the former first. Minor abdominal wall defects can be repaired after correction of the intracardiac defects.

This case report stresses the importance of early recognition of this rare disease and early referral to appropriate cardiac centre, so medical treatment followed by surgical repair can be accomplished at appropriate time.

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