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Pentalogy of Cantrell: the first Maltese case with successful outcome

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Introduction

Pentalogy of Cantrell is a rare disorder which was first described by Cantrell and his colleagues in 1958[1]. It is comprised of congenital heart disease (CHD), and midline defects. The latter consist of a supra-umbilical abdominal wall defect, a defect of the lower sternum, deficiency of the anterior diaphragm and a defect in the diaphragmatic pericardium, all of which contribute to varying degrees of ectopia cordis. Synonyms for this condition include Cantrell Pentalogy, Cantrell Syndrome, Cantrell-Haller-Ravich Syndrome, Pentalogy Syndrome, Peritoneopericardial Diaphragmatic Hernia, Thoracoabdominal Syndrome (TAS), TAS Midline Defect, and Thoracoabdominal Ectopia Cordis.

We report the first Maltese case which successfully underwent a two-stage repair for her CHD and ectopia cordis, and review the literature on this condition.

Patient

A female infant was born in January 1996 by normal vaginal delivery at full term to healthy and unrelated parents. This was the first product of this marriage and there was no family history of CHD or other malformations. At delivery, the apex of the heart was noted to be protruding through an epigastric hernia and visibly pulsating, underneath a thin layer of pericardium and skin (partial ectopia cordis). The hernia had two major components. The upper, smaller component contained the cardiac apex, while the lower, larger component contained bowel. The sternum was short (Figure 1).

Echocardiography on the same day showed mesocardia (midline heart), a large perimembranous ventricular septal defect (VSD), a large atrial septal defect (ASD) and a patent ductus arteriosus. Screening ultrasound examinations of the brain and abdomen were normal.

Initial treatment in Malta included diuretics for heart failure, and the exomphalos was covered with dry dressings.

At the age of 9 months, weighing 6.6 kg, the patient underwent corrective surgery in the UK whereby a Goretex patch was applied to the VSD and the ASD was directly sutured. The ductus arteriousus had closed spontaneously. The post-operative course was uneventful, and abdominal repair of the exomphalos was carried out two weeks after cardiac surgery. A 2 by 2 cm defect in the anterior part of the diaphragm was directly sutured, and the abdominal wall defect was closed by approximating the abdominal musculature.

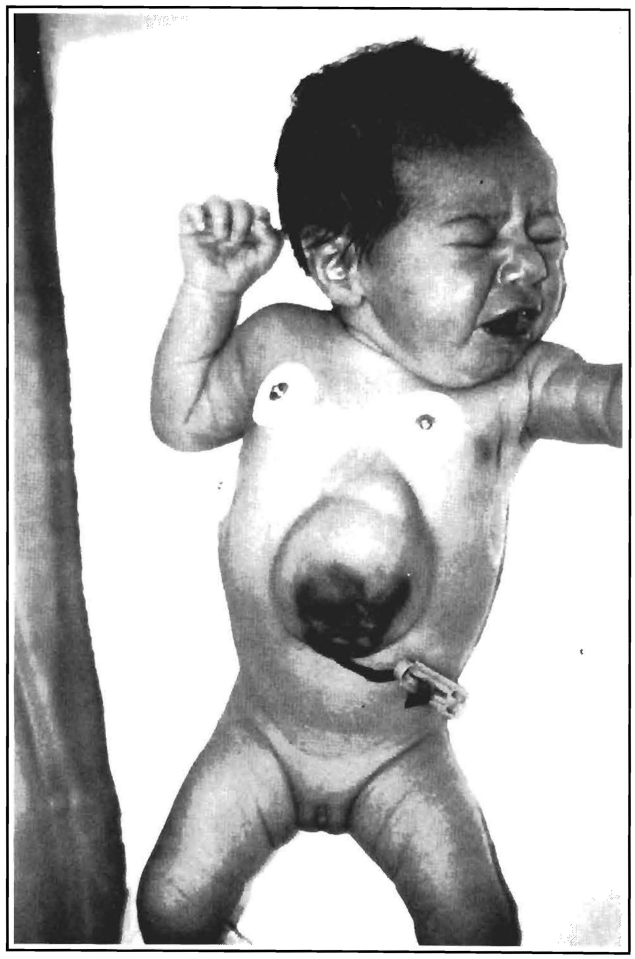

Figure 1: Cantrell's pentalogy. The abdominal hernia has two major components: the upper, smaller component contains the cardiac apex; the lower, larger component contains bowel.

Since then, the patient has developed normally and remains asymptomatic. Echocardiography shows no residual cardiac defects, and the abdominal wall repair is intact.

Discussion

The first cardiac malformation ever described was ectopia cordis. This description dates to 4000 BC in a clay tablet from the Royal Library of Nineveh[2]. Ectopia cordis was associated with calamities for the entire country.

Cantrell's pentalogy is a rare condition occurring in 5.5/million live births[3]. It is usually sporadic but one family had three consecutively born brothers with extensive diaphragmatic defects. Two of these siblings had the full pentalogy[4]. Antenatal diagnosis by means of ultrasound scanning is possible[5]. The commonest cardiac lesions are ventricular septal defect and tetralogy of Fallot.

Few cases have been reported since Cantrell described this condition and only a small proportion have survived[5]. Poor survival is due to the variable and often complex intracardiac malformations found in these infants[6], along with other malformations associated with this condition. These include pulmonary hypoplasia[6,7], left ventricular diverticulum[8], syrenomelia (fusion of lower limbs into one extremity)[9], additional central diaphragmatic herniation[10], cleft lip and palate[3], encephalocele[3] and fatal syndromes such as Edward's syndrome[11]. However, in a recent series, a survival rate of 50% among 10 treated patients with Cantrell's pentalogy with significant CHD was reported[6]. These included patients with absent right or left ventricular chambers in whom surgical repair culminated in a single ventricle physiology.

Repair is usually two-stage with separate operations for the thoraco-abdominal defects and cardiac malformation/s[6]. Single stage repair has also been successfully carried out[12], but multi-stage intracardiac repair may be necessary for complex CHD[6].

Conclusion

Pentalogy of Cantrell is a complex, midline developmental anomaly involving various organ systems. Surgical treatment is possible with internalisation of the heart, repair of associated intracardiac anomalies and repair of the thoraco-abdominal defect. Poor outcome is related to the presence of pulmonary hypoplasia (if any) and the severity of associated CHD.

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