

An Incomplete Pentalogy of Cantrell

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Pentalogy of Cantrell is a rare entity of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium and heart. The complete pentalogy and its variants have been described in the literature. We report a 47-day-old girl with an incomplete pentalogy of Cantrell presenting with congenital absence of the sternum, congenital heart defects, and an epigastric hernia. Two even rarer expressions of this disorder, tricuspid atresia type IIc and asymmetric kidneys, were also found. The patient was successfully managed with palliative pulmonary artery banding as preparation for a modified Fontan operation when she is older. We report this rare case, and discuss the pathologic findings and surgical strategy. (*Chang Gung Med J* 2008;31:309-13)

Key words: asymmetric kidneys, pentalogy of Cantrell, tricuspid atresia

Pentalogy of Cantrell is a rare entity of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium and the heart.⁽¹⁾ The complete pentalogy and its variants have been described in the literature.^(1,2) In this report, we present a case of an incomplete pentalogy with additional renal involvement.

CASE REPORT

A 47-day-old Palestinian girl manifesting symptoms of pulmonary hypertension was transferred to this hospital for surgical treatment of complex heart defects. In the local hospital, a diagnosis of congenital absence of the sternum, complex congenital heart defects (transposition of the great arteries with ventricular and atrial septal defects, and tricuspid atresia without pulmonic stenosis), and epigastric hernia had been established. She was acyanotic. On admission, physical examination revealed a blood pressure of 108/86 mmHg, a heart rate of 185 beats/min, and a body temperature of 37.9°C. She was malnourished with a body length of 45 cm, and weight of 3.5 kg.

She had a depression in the central portion of the chest wall with gross movements on respiration. The movements of the heart were visible. Sternal bony tissue could be palpated only in the upper portion of the chest wall. The heart could be palpated directly under the skin between the medial ends of the ribs. Auscultation of the chest revealed a grade 4/6 ejection systolic murmur that was loudest along the left sternal border, and was accompanied by a systolic thrill. Examination of the abdominal wall showed a "bubble" visible under the skin of the baby's belly between the umbilicus and xiphisternum when she cried, which disappeared when she was quiet. No other abnormalities of the head, face or limbs were noted. A chest roentgenogram in the frontal view showed generalized cardiomegaly and increased pulmonary vasculature, with a normal diaphragm. Preoperative echocardiography identified details of the congenital heart defects, including two ventricular septal defects, one large atrial septal defect with a right-to-left shunt, in addition to tricuspid atresia and transposition of the great arteries (Fig. 1). Nevertheless, the complex congenital heart defects

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could be defined as tricuspid atresia type IIc. Ultrasonography showed no evidence of a cranial or diaphragmatic abnormality. Distension of the hepatic veins and a disparity between the kidneys, with the

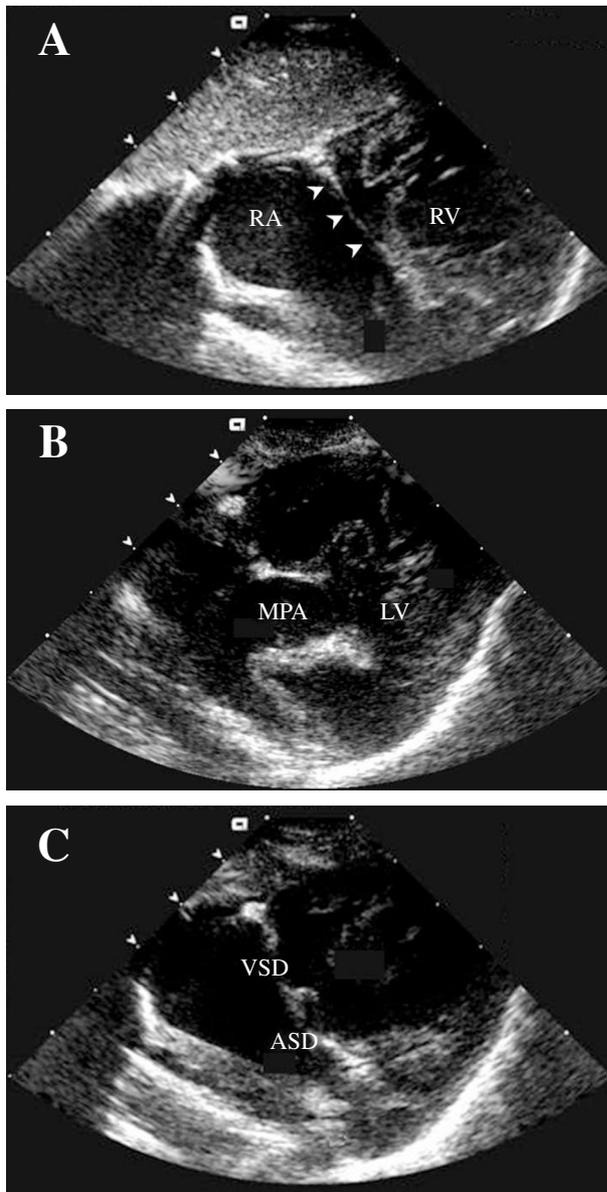


Fig. 1 Echocardiography of the congenital heart defects. (A) An apical view showing an echogenic atretic tricuspid valve (arrow heads); (B) A long-axis view showing a large pulmonary artery arising from the enlarged left ventricle; (C) A four-chamber view showing atrial and ventricular septal defects. ASD: atrial septal defect; LV: left ventricle; MPA: main pulmonary artery; RA: right atrium; RV: right ventricle; VSD: ventricular septal defect.

right longer than the left (Fig. 2), were noted. A diagnosis of an incomplete pentalogy of Cantrell was established.

Pulmonary artery banding was performed two

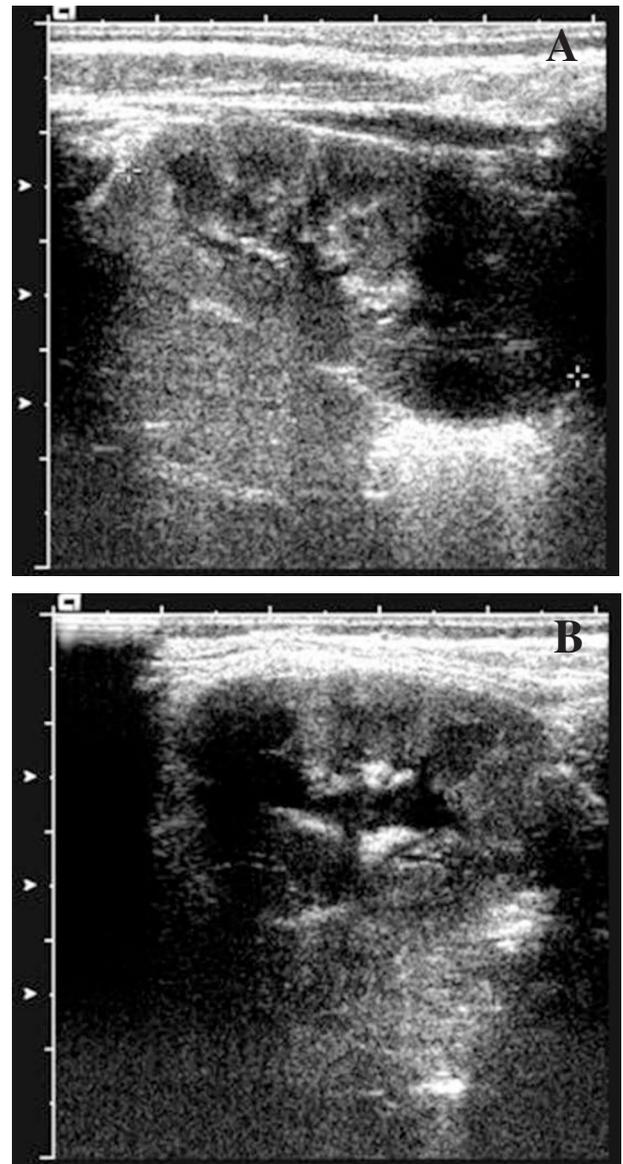


Fig. 2 Longitudinal gray scale ultrasound images of (A) the right and (B) the left kidneys. The right kidney is much longer than the left. The kidneys are otherwise normal without hydronephrosis. The right kidney has a hyperechoic renal cortex, and the renal sinus is concentrated centrally. Although the left kidney is shorter, it is a little larger in the short axis than the right. The renal cortex is more hypoechoic, the renal sinus is more scattered, and the medullary pyramids are larger than in the right.

days later. The operation was carried out with the patient in a supine position. During surgery only the manubrium was found instead of the sternum. It was 1.7 cm long, with the lower portion missing and occupied by fibrotic tissue, and it was fused with the intact frontal pericardium. The anterior ends of the ribs were visible in the normal position. A sternotomy was made on the manubrium, and a retractor was applied between the manubrium and ribs. The pericardium was intact. No diaphragmatic hernia was noted after the pericardium was opened. The main pathologic findings in the cardiovascular system included a generalized cardiomegaly, and reversed aorta and pulmonary artery trunk. The anteriorly placed small aorta arose from a small right ventricle, while the posteriorly placed enlarged pulmonary artery arose from an enlarged left ventricle.

Since total correction was not being done at this time, a band was placed on the pulmonary artery after it was mobilized, and the systolic pulmonary artery pressure decreased from 90 mmHg to 30 mmHg. The manubrium was sutured with a single wire, and the pericardium was closed from bottom to top. After the operation, the patient's condition improved progressively. Her postoperative course was uncomplicated. She required only minimal inotropic support for 24 hours and was extubated on the first postoperative day. She remained in sinus rhythm and was continued on a regimen of digoxin and diuretics. She did not manifest symptoms of significant heart failure. A week after surgery, a chest roentgenogram showed no obvious reduction of heart size or pulmonary vasculature. She was discharged on the 9th postoperative day.

DISCUSSION

Pentalogy of Cantrell is a rare entity of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium and heart.⁽¹⁾ It was first reported by Cantrell *et al.*⁽¹⁾ in 1958. The incidence has been estimated to be 1 in 65,000-100,000 live births.^(3,4) The syndrome occurs with various degrees of severity from incomplete to severe expression with involvement of other organ systems. The complete syndrome with all five defects has been documented, while presentation with four of the five defects including an intracardiac lesion and ventral abdominal wall defect is regarded as a probable syn-

drome.⁽²⁾ All five defects plus additional abnormalities or more extensive involvement have been seen in severe cases.⁽²⁾ Toyama reported eight patients with the incomplete syndrome.⁽²⁾ Bhat *et al.*⁽³⁾ reported a case of incomplete pentalogy of Cantrell including absence of the sternum, a large omphalocele, and heart abnormalities, and called this disorder Cantrell syndrome. The etiology of the pentalogy is not well established. A widely-accepted theory, which was proposed by Cantrell *et al.*,⁽¹⁾ stated that developmental failure of the mesoderm in early embryonic life between 14 and 18 days of gestation results in a failure in the development of the transverse septum of the diaphragm, and of the ventromedial migration of the paired mesodermal folds of the upper abdomen. Congenital defects of the sternum may vary from simple notching of the manubrium and irregularities in the shape of the xiphoid to absence of the entire sternum.⁽⁵⁾ Abdominal wall defects include omphalocele, diastasis recti, epigastric hernia, umbilical hernia, and combined defects. The most common abdominal wall defect is omphalocele.⁽³⁾ Deficiencies of the diaphragmatic pericardium and the anterior diaphragm are common defects. Sometimes, they are too small to be noted.⁽⁶⁾ Cardiac lesions may vary widely. Cantrell *et al.*⁽¹⁾ stated that congenital intracardiac anomalies are consistent elements of the pentalogy, with ventricular septal defect in every case (100%), atrial septal defect in 53%, pulmonary stenosis in 33%, tetralogy of Fallot in 20% and left ventricular diverticulum in 20%. Since 1958, a few reported cases have included cyanotic tricuspid atresia, some of which were successfully managed with a palliative modified Blalock-Taussig shunt.^(2,7-9) However, acyanotic tricuspid atresia type IIc has not been previously described in this disorder.

In a collective review by Toyama,⁽²⁾ including 36 cases of pentalogy of Cantrell reported from 1772 to 1970, additional defects included head and facial deformities, meningocele, anencephalia, cleft lip, cleft palate, lung hypoplasia, adrenal aplasia, malrotation of the colon, hernia of the bowel into the pericardial cavity, undescended testicle, and deformities of finger and foot. Renal involvement is a rarer additional defect in the pentalogy. Fernández *et al.*⁽¹⁰⁾ reported 4 cases of pentalogy of Cantrell, with one patient having agenesis of the right kidney. Pollio *et al.*⁽¹¹⁾ reported three patients with the syndrome, one of which showed had a dysplastic left kidney and

mild pyelectasis of the right kidney. Left renal agenesis was found by Aslan *et al.*⁽¹²⁾ in an autopsy of a patient with pentalogy of Cantrell who only survived 15 minutes. Assessment of asymmetric kidneys has led to the conclusion that an abnormally small kidney is morphologically normal but has either a reduced number of nephrons or small nephrons. Blood tests of renal function would not be affected unless 70%-75% of the kidney is nonfunctioning. This phenomenon is considered to be due to cessation of embryonic development of the kidney, the structure of which is otherwise normal. The cause is not yet clear and could be related to an abnormal synthesis of growth factors or their receptors during the sensitive phase of metanephrogenesis.⁽¹³⁾ It is evident that the disparity of the kidneys in the present patient was an additional expression of the pentalogy.

The only known case of a hepatic lesion in pentalogy of Cantrell was reported by Aslan *et al.*,⁽¹²⁾ who noted adreno-hepatic fusion in one of their patients. A pure distension of the hepatic vein in the present case was more likely to be associated with congestive right-sided heart failure than a hepatic defect from the pentalogy.

Toyama⁽²⁾ demonstrated a survival rate of 20% in this disorder including its variants and incomplete syndromes. The complete pentalogy has a poorer outcome, and the survival rate was only 5/59 (8.5%) in the report of Fernández *et al.*⁽¹⁰⁾ There is a general agreement that pentalogy of Cantrell is lethal without surgery, and successful corrective or palliative operations have been done, especially in less severe cases.⁽⁶⁾ With incomplete pentalogy of Cantrell, even with additional kidney involvement, the present patient has an encouraging prognosis. However, the opportune moment for further correction remains controversial because of the underlying cardiac and sternal defects. It has been proposed that the sternal defect be reconstructed early as problems in management increase considerably with advancing age.⁽¹⁴⁾ Pulmonary artery banding has already been done as an interim therapeutic strategy. Definitive repair of the cardiac defect has to be postponed until the baby grows to a suitable age and body weight, with a proper pulmonary artery pressure. The age of patients in modified Fontan operations has decreased from 11.8 years in the early years of treatment to 2.5 years by the end of the last century, with a low early

mortality of 4-0%.⁽¹⁵⁾ Hence, a modified Fontan operation could be performed when she reaches 2.5 years old. This might be followed by a sternal reconstruction and epigastric hernia repair. It is noteworthy that the size and nature of the sternal defect are the determinants of the closure technique.⁽¹⁴⁾ The reconstructive techniques and materials of choice include prosthetic grafts (Marlex mesh, Teflon, silicone elastomer, or perforated acrylic plate), homografts (flaps of bone, fascia lata, or homograft rib), and autografts (autologous ribs or cartilage, pectoris major muscle).^(5,14,16) A pectoralis major muscle would be preferable in providing sufficient strength and stability, and avoiding potential infections.

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