

Case Report

Incomplete pentalogy of Cantrell: a case report

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ABSTRACT

Background: Pentalogy of Cantrell is a rare congenital syndrome characterized by defects involving the supra-umbilical abdominal wall, lower sternum, anterior diaphragm, pericardium, and the intra-cardiac anomalies. The ensuing congenital heart disease is a source of major morbidity. The full spectrum of the syndrome is rare while incomplete expression of the syndrome and its variants are well-recognized.

Case presentation: A 6-day-old male neonate with an incomplete form of Cantrell pentad was reported. At 22 weeks' gestation, prenatal sonography indicated omphalocele and ventricular septal defect (VSD) with a double outlet right ventricle (DORV). The patient was referred to Khon Kaen University Hospital (Srinagarind Hospital) in Northeast Thailand. Amniocentesis revealed 46,XY. He was born full-term by normal delivery. His congenital defects were confirmed; however, he died 2 days after surgical correction of the abdominal wall and diaphragm defects. The distinct findings relevant to pentad were noted.

Keywords: Incomplete pentalogy of Cantrell, diaphragmatic hernia, double outlet right ventricle, omphalocele

INTRODUCTION

Pentalogy of Cantrell is a rare disease, with multiple congenital thoraco-abdominal malformations (1) a midline supraumbilical abdominal wall defect; (2) a defect of the lower sternum; (3) a defect of the diaphragmatic pericardium; (4) a deficiency of the anterior diaphragm, and (5) congenital cardiac anomalies¹.

It was first described by Cantrell et al. in 1958¹. The incidence of the syndrome is about 1 in 65,000 live births². The condition is believed to be caused by a failure in development of the lateral mesoderm; which occurs when the embryo is about 14-18 days old, when the splanchnic and somatic mesoderm is dividing and developing¹. The failure of the lateral mesodermal folds to the midline

resulting in sternal and abdominal defects while failure of the septum transversum to develop causes defects in the anterior diaphragm and peri-cardium as well as abnormal development of the myocardium from the splanchnic mesoderm¹. Organs may eviscerate through the resulting sternal and abdominal wall defects (Figure 1), making prenatal diagnosis of the syndrome feasible by ultrasonography. The full spectrum of pentalogy is a rare occurrence while the incomplete expression of the syndrome; the combinations of two or three organ defects included in the pentad with either intracardiac and abdominal wall defects or a sternal abnormality³, and its variants are well-recognized^{3,4,5}. Pathogenesis trisomy 13, and 18 has not been fully elucidated. It occurs sporadically, but has also been recorded in twins^{6,7} and in patients with trisomies 13, 18 and Turner syndrome^{8,9}. Herein we report an incomplete form of the syndrome in a neonate. The distinct findings relevant to pentad—including

the congenital heart anomalies similar to TOF—are highlighted.

CASE PRESENTATION

A male, full-term infant was born by normal delivery to a 25-year-old woman (gravida 2, para 1). The child was the second for the parents; the first gravida was aborted for an unknown cause at 15 weeks' gestation. The mother denied any exposure to known teratogenic agents. The parents were not consanguineous. The infant underwent prenatal sonography at 22 weeks: the diagnosis was omphalocele and intracardiac anomalies (VSD with DORV), so was transferred in his last trimester to Srinagarind Hospital for antenatal care. The pregnancy proceeded to term even after amniocentesis revealed the 46,XY karyotype. At birth, the infant had central cyanosis and an abdominal wall defect with omphalocele (5 cm), containing a partial extra-corporeal liver (Figure 1A). The abnormal

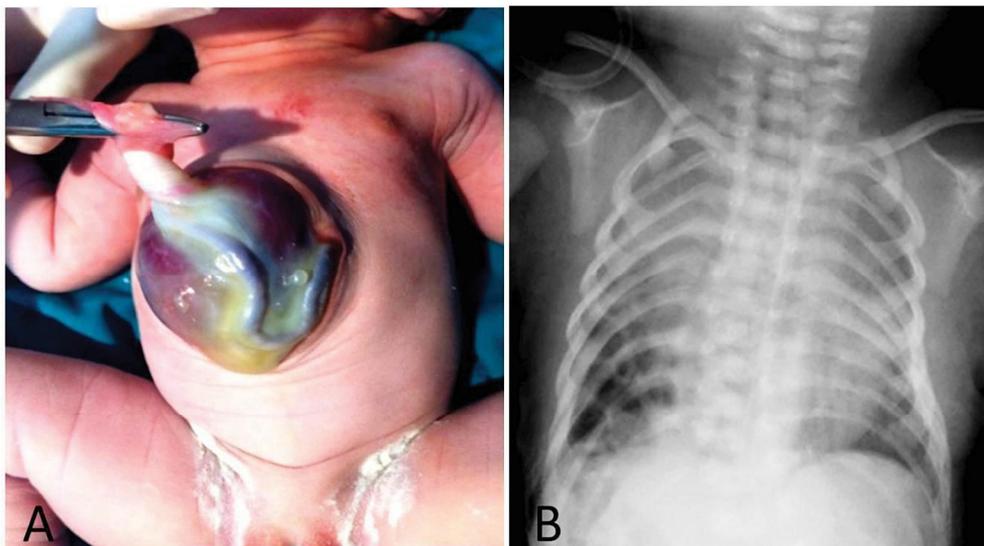


Figure 1 Infant with incomplete form of pentalogy of Cantrell: (A) supraumbilical abdominal wall defect—an omphalocele, containing extracorporeal liver with an umbilical cord insertion at the sac near the upper margin of the defect; (B) diaphragmatic defect—plain film of chest showing bowel gas pattern at lower right lung field with displacement of cardiac shadow

heart sound was a SEM grade II at LLPSB. A chest X-ray revealed bowel gas at the right lung field (Figure 1B) and ultrasound demonstrated a small defect at the anterior part of the diaphragm. Echocardiography suggested tetralogy of Fallot (TOF) anomalies and the differential diagnoses included DORV with VSD. The infant underwent surgery to repair the diaphragmatic hernia and close the omphalocele. The patient died two days after surgery.

PATHOLOGICAL FINDINGS

The pathological findings revealed surgical wounds repairing two of the pentad defects at mid abdomen (8 cm) (repairing the omphalocele) and a diaphragmatic hernia wound at the anterior part of the diaphragm. Congenital cardiac anomalies comprised: (a) a patent ductus arteriosus (PDA, 5x18 mm, type C-long tubular without constriction),

(Figure 2A); (b) a secundum atrial septum defect (ASD, 5 mm); (c) hypertrophy of right ventricle (9 mm thick wall); (d) DORV with aortic valve and pulmonary valve (15 and 12 mm circumference, respectively); and, (e) subaortic-, perimembranous VSD (7 mm) associated with anterior malalignment of the muscular outlet septum (abnormal anterior insertion of supra-ventricular crest into anterior limb of septomarginal trabeculation) causing narrowed subpulmonic infundibulum (TOF- type DORV) (Figure 2B). This arrangement, with obstruction of the ventricular outflow tracts, resembles to physiology of TOF.

DISCUSSION

We described a neonate with a normal karyotype, diagnosed before birth as an incomplete pentalogy of Cantrell with complex intracardiac defects. The infant died on the sixth postnatal day,

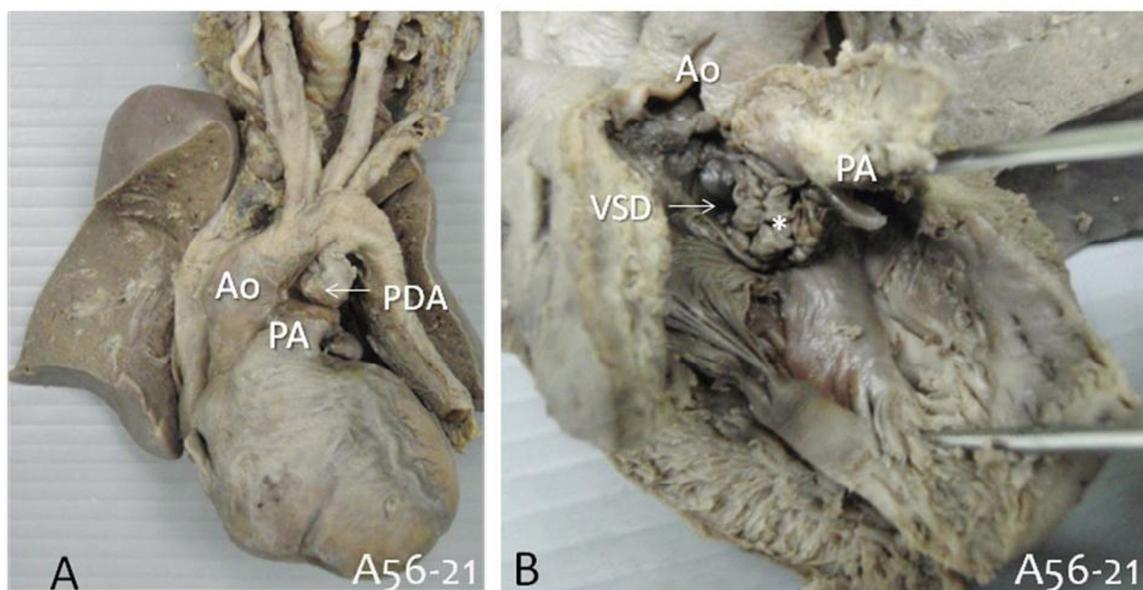


Figure 2 Congenital cardiac defects: (A) PDA (arrow)—type C (long tubular type); (B) right ventricular view showing ventricular hypertrophy, double outlet right ventricle with subaortic VSD (arrow) and the anterior deviation of the muscular outlet septum (*). Ao, aorta; PA, pulmonary artery; PDA, patent ductus arteriosus; VSD, ventricular septal defect.

2 days after surgical intervention to repair the abdominal wall, and diaphragm defects. The pathological findings confirmed the diagnosis, as we found evidences of abdominal wall and diaphragmatic defects and intracardiac anomalies (TOF-type DORV). A literature review of patients with pentalogy of Cantrell (33 cases with complete form and 23 cases incomplete form), the following types of congenital heart disease were noted: VSD (12/33 and 8/23 cases); ASD (7/23 and 5/23 cases) and TOF (6/33 and 3/23 cases), DORV (0/33 and 3/33 cases)¹⁰. Among these 56 patients various other associated anomalies were reported, including: (a) craniofacial and central nervous system anomalies (cleft lip and/or palate, encephalocele, and hydrocephalus) in 13/56 cases; and, (b) limb defects (club-foot, absence of tibia or radius, and hypodactyly) in 10/56 cases¹⁰. Associated intrabdominal anomaly is uncommon and found in only 1/56 cases (agenesis of the gall bladder, and polysplenia)¹⁰. This report supports the proposal by Carmi and Boughman[5] that pentalogy of Cantrell should be included among the defects of the ventral midline developmental field rather than abnormal segmental development, as previously suggested by Cantrell¹. Prenatal detection of an omphalocele, associated with multiple midline defects, should alert the practitioner to the possibility of pentalogy of Cantrell and also OEIS complex¹¹. The OEIS complex is characterized by a combination of omphalocele, exstrophy of the bladder, an imperforate anus, and spinal defects. Cytogenetic analysis is necessary at the time of prenatal diagnosis of pentalogy of Cantrell. Families tend to terminate affected pregnancies because of the intracardiac defects associated with this syndrome, which can reduce post-natal and post-surgical survival^{11,12}. Nevertheless, successful two-stage repair of the TOF (associated with pentalogy of Cantrell) has been reported¹³, and one-stage repair of

pentalogy of Cantrell is technically feasible¹⁴. Some children with mild thoraco-abdominal abnormalities have survived into adulthood¹⁵. The information of chromosome analysis is useful in family counseling and can influence perinatal management of affected fetuses. Importantly, pentalogy of Cantrell has a negligible recurrence risk¹⁶.

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