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Case Report

Baby girl with pentalogy of Cantrell: a case report on an extremely rare condition

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In 1958, Cantrell *et al.* described an extremely rare syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium, and heart malformation.[1] The incidence of pentalogy of Cantrell (POC) is one in 65,000 live births.[1,2,3] Only one case was reported in Dr. Moewardi Public Hospital between 1999 and 2016. The exact etiology of this condition is unknown, but developmental failure of mesoderm at 4 weeks of gestational age may contribute to the condition.[4] The prognosis depends on the degree of heart failure and the malformations that occur. Heart failure, arrhythmia, cardiac rupture, cardiac tamponade, endocarditis, and peripheral emboli are described as the main complications and causes of death.[5,6] The aim of this report was to add to reference data about complete POC and the prognostic outcome. **[Paediatr Indones. 2019;59:51-4; doi: http://dx.doi.org/10.14238/pi59.1.2019.51-4].**

Keywords: *pentalogy of Cantrell; ectopia cordis; omphalocele*

The Case

A baby girl weighing 2,800 grams was born full term, cried spontaneously and had no cyanosis. Her mother was 20 year old and this was her first pregnancy. The baby was admitted to our hospital due to a big lump on her abdominal wall. There was no family history of congenital defects. At birth, the baby's vital signs were normal and the ictus cordis (the apex beat) appeared normal with regular heart sounds. Physical examination showed a palpable soft mass resembling omphalocele (4x5x6cm) with ectopia cordis on the supraumbilical defect (**Figure 1**). Electrocardiography demonstrated sinus rhythm with left ventricular hypertrophy and echocardiogram showed defects of the subaortic ventricular septum, left-to-right shunt, and moderate pulmonal stenosis with 55% ejection fraction (**Figure 2**).

Figure 1. A palpable soft mass resembles omphalocele (4x5x6cm) with ectopia cordis on the s praumbilical defect.

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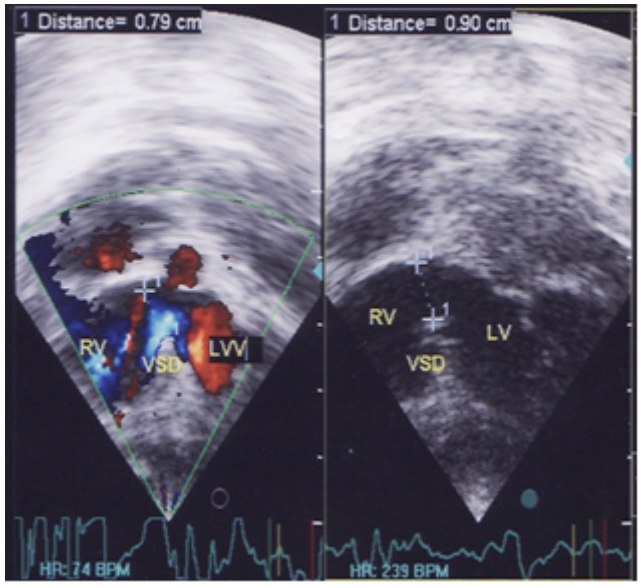
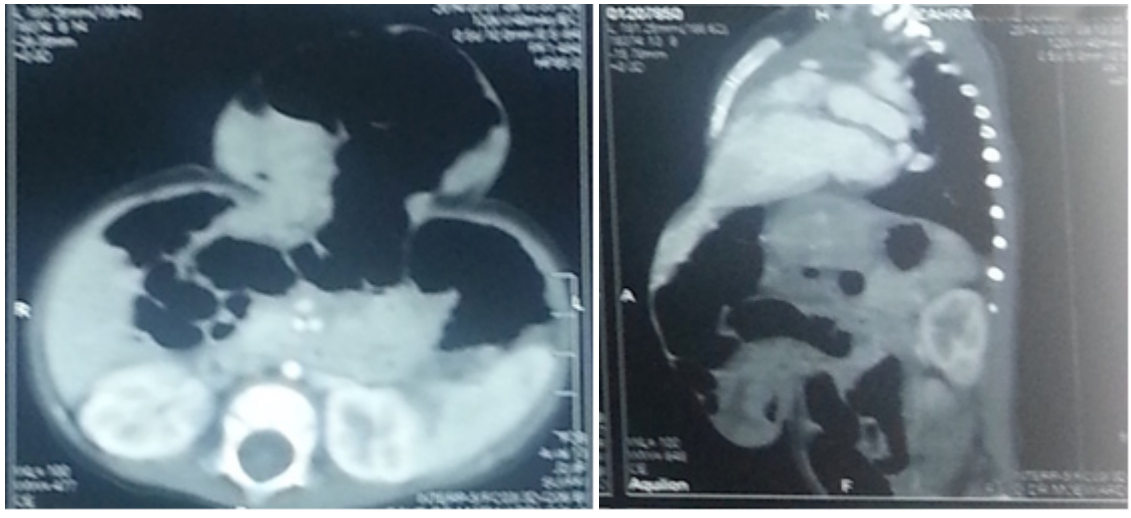


Figure 2. Electrocardiography demonstrated sinus rhythm with left ventricular hypertrophy, while echocardiography showed subaortic ventricular septum defect, left-to-right shunt, and moderate pulmonal stenosis with ejection fraction 55%.

The thoraco-abdominal CT scan with contrast was performed due to the cardiac abnormalities cited above and it revealed a huge cardiac diverticulum starting from the left ventricular apex to the umbilicus, accompanied by omphalocele. Supraumbilical defects followed by a defect in the lower sternum of 62.1 mm in diameter (**Figures 3a and 3b**).

The left ventriculography catheterization revealed adequate right ventricle size and a flow from the right ventricle to the pulmonary artery. Right ventriculography revealed confluent pulmonary artery, visible left diaphragmatic hernia with intestine in front of the left lung, and heart protruding forward. The left ventricle had a visible ventricular septal



Figures 3 (a. axial view, b. sagital view). The contrasted thoracoabdominal CT scan showed asserting cardiac abnormalities and a huge cardiac diverticulum starting from the left ventricular apex to the umbilicus, accompanied by omphalocele as well as supraumbilical defects with the defect in the lower sternum of 62.1 mm in diameter.

defect with left-to-right shunt. Aortography showed no collateralization and no patent ductus arteriosus. During the catheterization, the patient had ventricular tachycardia and died in the pediatric intensive care unit due to heart failure.

Discussion

Cantrell et al. first described the full spectrum of POC in 1958.[1] Cantrell's pentad, it is an extremely rare congenital malformation and usually lethal, with multiple defects in the midline supraumbilical abdominal wall, anterior diaphragm, sternum, as well as diaphragmatic, pericardial, and heart defects.[1-5] Complete POC is rarely reported. Thus, we report a case of complete POC, with hopes of improving our knowledge on this extremely rare congenital condition.

Toyama described the following classifications of POC: Class 1- complete syndrome with all five defects present; Class 2- probable syndrome, with four defects present, including intracardiac and ventral abdominal wall abnormalities; and Class 3- incomplete syndrome with various combinations of defects present, always with sternal abnormalities.[6] The failure of pathophysiology and embryologic development during pregnancy results in various abnormalities, especially those derived from the mesoderm layer.1-5 The extracardiac disorders include omphalocele, anterior diaphragmatic defects, sternal defect, may also be observed in association with ectopia cordis.[7]

Omphalocele should be considered pathologic only, if it persists beyond 14 weeks or if its maximum diameter exceeds 1 cm in the first trimester of pregnancy.[8] The sternal and abdominal wall defects represent faulty migration of these mesodermal primordial structures. It is thought that these developmental abnormalities occur from approximately days 14 to 18 of embryonic life.[8] The anterior abdominal wall defects are mostly omphalocele. A retrograde ventral diaphragm defect occurs in 91% of cases. Mortality is higher in infants with the complete form of POC and associated with extracardiac anomalies.[9] In the literature, thoraco-abdominal ectopia cordis (EC) is the usual type of ectopia cordis associated with Cantrell's pentalogy. Ectopia cordis is defined as an abnormal location of the heart, partially or totally outside of the thorax. Both are in the cervical, cervicothoracic, chest, and thoracoabdominal regions.[10] The incidence of ectopia cordis is 5.5-7.9 per one million births, and only about 0.5 to 0.8% of all congenital heart diseases.[7] Ectopia cordis is mostly associated with other congenital heart anomalies, including ventricular septal defect, atrial septal defect, pulmonary stenosis, Tetralogy of Fallot, and right ventricular diverticulum.[7,10] Mortality in children with EC is high; no more than 5% survive.[9] The POC usually can be diagnosed in the first trimester of pregnancy with prenatal ultrasonography, usually made by 2-dimensional ultrasound examination, whereas 3-dimensional ultrasonography is more useful in making a diagnosis in the second and third trimesters.[10,11]

In our case, the sternal defects were the absence of xiphoideus and the lower 2/3 of the sternum. Cardiac rupture, tamponade, sudden death, endocarditis, peripheral embolism, heart failure, and arrhythmia have all been described as complications and causes of death. Sternal defects include bifid sternum (26%), absent xiphoid (10%), and absent lower 2/3 of sternum (9%). A ventral retrosternal defect of the diaphragm occurs in 91% of the cases.[11]

In conclusion, the pathogenesis of POC is uncertain. It arises from an embryological development defect occurring in a segment of lateral mesoderm, 14-18 days after conception. The survival rate for patients with complete POC is low, with the prognosis depending on the severity of the cardiac defect and complexity of the malformations.

Conflict of Interest

None declared.

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