INTRODUCTION

The Pentalogy of Cantrell (PC) usually presents at birth. Incidence of the PC varies from 5.5 to 7.9 per million live births. PC is a collection of five congenital midline birth anomalies, which included a defect of the lower sternum, a midline supraumbilical thoracoabdominal wall defect, a deficiency of the diaphragmatic pericardium, a deficiency of the anterior diaphragm, and congenital cardiac anomalies. The pathognomonic characteristic of this syndrome is the coexistence of omphalocele and ectopia cordis. Diagnosis is possible by antenatal or physical examination, and other tests should be performed to detect other anomalies, including the heart's anomaly. Surgical treatment is complex, and various operative techniques often require a staged approach. The mortality rate is quite high, with a survival rate of less than 5%.

Case presentation: Two-day-old male infant was complained that his heart had been outside his chest wall since birth without covering by skin or sternum. He was born on a term through section cesarean with a vigorous baby condition. Section cesarean was chosen because his mother has Human Immunodeficiency Virus (HIV)—no family history with similar symptoms. While pregnant, the mother regularly checks with the midwife and fetal ultrasonography only once during the third trimester but did not find the defect, no history of drug abuse, teratogen exposure, smoking, alcohol, or consanguinity. The midline defect extended from the lower part of the sternum completely. Echocardiography found defects such as classic Tetralogy of Fallot, mild atrial regurgitation, mild mitral regurgitation, mild tricuspid regurgitation, and suspect mass at left atrium (LA). The patients underwent surgery by covering the heart with a sile bag. Three days after the procedure, the patient's condition worsened and death because of the sepsis.

Conclusion: Prenatal diagnosis is important in guiding the surgical team to estimate the prognosis and reduce a postnatal management strategy. Early management can provide the basic care necessary to keep the patients until definitive management can be provided.

ABSTRACT

Background: Pentalogy of Cantrell (PC) is a rare anomaly, and the incidence varies from 5.5 to 7.9 per million live births. The components are defined as a defect of the lower sternum, a midline supraumbilical thoracoabdominal wall defect, a deficiency of the diaphragmatic pericardium, a deficiency of the anterior diaphragm, and congenital cardiac anomalies. The pathognomonic characteristic of this syndrome is the coexistence of omphalocele and ectopia cordis. Diagnosis is possible by antenatal or physical examination, and other tests should be performed to detect other anomalies, including the heart's anomaly. Surgical treatment is complex, and various operative techniques often require a staged approach. The mortality rate is quite high, with a survival rate of less than 5%.

Case presentation: Two-day-old male infant was referred to our Hospital; Universitas Udayana/ Prof. Dr. IGNG Ngoerah Hospital. While pregnant, the mother regularly checks with the midwife and fetal ultrasonography only once during the third trimester but did not find the defect, no history of drug abuse, teratogen exposure, smoking, alcohol, or consanguinity. The midline defect extended from the lower part of the sternum completely. Echocardiography found defects such as classic Tetralogy of Fallot, mild atrial regurgitation, mild mitral regurgitation, mild tricuspid regurgitation, and suspect mass at left atrium (LA). The patients underwent surgery by covering the heart with a sile bag. Three days after the procedure, the patient's condition worsened and death because of the sepsis.

Conclusion: Prenatal diagnosis is important in guiding the surgical team to estimate the prognosis and reduce a postnatal management strategy. Early management can provide the basic care necessary to keep the patients until definitive management can be provided.

Keywords: pentalogy of Cantrell, ectopic cords, infant. 


Prenatal diagnosis is important in guiding the surgical team to estimate the prognosis and reduce complications and causes of death. This study reported a PC on a two-day-old male infant in Bali.

CASE

A two-day-old male infant was referred from Negara Hospital, Bali, on December
CASE REPORT

3rd 2021. He was complained that his heart had been outside his chest wall since birth without covering by skin or sternum (Figure 1). He was born on aterm through section cesarean with the vigorous baby condition. Section cesarean was chosen because his mother has Human Immunodeficiency Virus (HIV). The birth weight was 3150 grams, and the body length was 48 cm. He is the youngest child in the family and the son of the third husband. There was no family history with a similar symptoms. His mother also had HIV and already taking antiretroviral (ARV) since 4 years ago, and his father has not been checkup for HIV status—his mother’s history of having a fifth child miscarriage. While pregnant, his mother regularly checked to the midwife and fetal ultrasonography only once in 3rd trimester. However, she did not find the defect, no history in the family with the same disorder, and no history of drug abuse, teratogen exposure, smoking, alcohol, or consanguinity.

The patient was active on physical examination and appeared generally in good condition. The vital sign such as pulse, breathing, and body temperature were normal, but oxygen saturation was 85%-90% with cyanotic extremities. The midline defect extended from the lower part of the sternum completely. We had difficulty hearing the heart sounds because the heart lies outside the thoracic cavity. The right atrium, left atrium, right ventricle, and left ventricle were exposed completely without skin or membrane or pericardium coverage. The breath sounds were normal, and there were no additional sounds of chest auscultation. The central nervous system was normal.

The patient was under neonatologist, pediatric cardiologist, cardiothoracic and vascular surgeon and pediatric allergy immunology. Additional examination such as complete blood count, septic marker, and echocardiography was done to examine the baby’s conditions. The complete blood count investigation revealed hemoglobin 11.90 g/dL, MCV 105.80 fl, MCH 36.5 pg, MCHC 34.5 g/dL, white blood cells count 20.89x10$^3$/µL, neutrophil %/# 77.6%/16.21, lymphocytes %/# 11.5/2.25, platelet count 227x10$^3$/µL, IT ratio 0.11, the result blood smear was normocytic normochromic anemia and relative lymphopenia, and the blood culture was no growth. The patient got the first-line antibiotics in the NICU. Thorax and abdominal X-ray showed a suspicion of pneumonia, which is not supported by the physical examination
result, so we did not treat it as pneumonia (Figure 2). Echocardiography was done by a pediatric cardiologist who found that he had some intracardiac defects such as classic Tetralogy of Fallot, mild atrial regurgitation, mild mitral regurgitation, mild tricuspid regurgitation, and suspect mass at LA (Figure 3 and 4).

The palliative surgery aimed to cover the exposed heart and topical treatment. The patient got zidovudine 4 mg/body weight/times every 12 hours for therapy prophylaxis HIV exposed infant, antibiotics, and propranolol 2 mg every 8 hours.

The patient was planning surgery therapy by covering the heart with a bovine pericardial sac, but we did not have the material. So, the patients underwent surgery by covering the heart with a silo bag (Figure 5). After three days of covering the heart, the patient’s condition worsened and death because of sepsis.

DISCUSSION

Pentalogy of Cantrell (PC) is a rare anomaly first described in 1958 by James R Cantrell et al. The components of the PC are defined: midline, upper abdominal wall disorder (e.g., omphalocele, gastroschisis); defect of the lower sternum (i.e., cleft sternum or absent sternum); anterior diaphragmatic defect (i.e., hypoplastic diaphragm, anterior diaphragmatic hernia); pericardial abnormality (e.g., ectopia cordis); congenital abnormalities of the heart (e.g., tetralogy of Fallot, ventricular septal defect, atrial septal defect). The pathognomonic characteristic of this syndrome is the coexistence of omphalocele and ectopia cordis.3

Ectopic cordis (EC) is described as the malposition of the heart, partially or completely outside the thorax. According to the position of the misplaced heart, ectopia cordis can be classified into five types: 1) cervical, in which the heart is located in the neck with a sternum that is usually intact; 2) thymocervical, in which the heart is partially in the cervical region but the upper portion of the sternum is split; 3) thoracic, in which the sternum is completely split or absent, and the heart lies partially or completely outside the thorax; 4) thoracoabdominal, which usually accompanies Cantrell’s syndrome; 5) abdominal, in which the heart passes through a defect in the diaphragm to enter the abdominal cavity.2,4 In this case, we found the type of ectopia cordis with a predominant thoracoabdominal component.

The diagnosis PC can be detected when prenatal, using ultrasonography to find the abnormality, so it can usually diagnose as early as 10 weeks of pregnancy. It is important to mention that although accurate prenatal diagnosis of this rare condition is essential for effective postnatal management, the timing of the prenatal diagnosis does not affect the outcome. The diagnosis made by physical examination and other tests should be performed to detect other anomalies, including the anomaly of the heart itself, such as echocardiography dan radiography for finding the abnormalities from the sternum and diaphragm.2,4

Echocardiography is an excellent diagnostic tool to describe the structural anatomy of the heart. The most common associated defect is a ventricular septal defect (VSD) and tetralogy of Fallot (TOF), followed by another frequent defect: ASD, left ventricular diverticulum, pulmonary stenosis or atresia, and other less frequent defects.1 In this case, we found that intrinsic anomalies of the heart detected by echocardiography are classic TOF, mild AR, mild TR.

Toyama classified PC into: class I-the occurrence of all 5 defects, class II- the occurrence of 4 defects with intracardiac and ventral abdominal wall abnormalities present; and class III-incomplete expression of the disorder showing various combinations of defects, although sternal anomalies are present.2 In this case, we found omphalocele, ectopia cordis, and intracardiac abnormalities, so he categorizes as pentalogy of Cantrell class II.

The exact cause of the PC is unknown. The cases are mostly sporadic. Many familial cases suggest genetic factors may play a role in this disorder’s development, such as chromosomal abnormalities like trisomy 21, trisomy 18, Turner syndrome, and other anomalies.3,7

Delivery should be ideally via cesarean section, as vaginal delivery can result in prolonged cardiac compression, damage to the herniated viscera, and rupture of an atrial diverticulum or the sac of the omphalocele.3

During the third week of gestational life, when the demarcation between the intraembryonic and extraembryonic coeloms is established, the development of the body cavities and diaphragms begins. By the beginning of the fourth week, a horseshoe-shaped cavity develops in the cardiogenic and lateral mesoderms. The curve of the horseshoe represents the future pericardial cavity. During this period, the pericardial and peritoneal cavities communicate dorsally through the pericardio-peritoneal canals. The developing septum transversum will ultimately separate the thoracic and abdominal cavities by forming the diaphragms—the fibrous pericardium forms from the pleuropericardial folds. The folds grow toward the midline and attach to the primitive mediastinum and end up separating the heart from the lungs, with the thoracic cavity now divided into the lungs, with the thoracic cavity now divided into a pericardial cavity and two pleural cavities. During the folding of the embryonic disc in week 4, the lateral body walls coverage together on the ventral aspect of the embryo and fuse. At this stage, incomplete fusion may occur and result in the partial or complete evisceration of the heart through the defect.9

Most prior studies examining the risk of congenital anomalies (CAs) according to in-utero ARV exposure have been reassuring. However, some evidence has suggested an increased risk CAs overall or for certain CAs with specific ARVs. In the International Antiretroviral Pregnancy Registry, the estimated prevalence of CAs was 2.9% among more than 6900 children with first-trimester ARV exposures, similar to the rate among children exposed in the later trimester.10,11 The infants exposed to HIV with a history of mother consumption of ART have a low risk for congenital disabilities. The benefits of recommended ARV use during pregnancy still outweigh such risks.11

Clinical presentations reported from several cases are focused on intrinsic cardiac anomaly and sepsis. Infants are mostly present with shortness of breath due to pumping mechanism disturbances due to associated intrinsic anomalies of
the heart. Fever is caused by the absence of skin and pericardium, thus making it susceptible to bacterial contamination and may lead to sepsis. Treating sepsis becomes difficult to manage as long as atmospheric contact is not repaired. Early surgical intervention may be a risk factor for mortality, and stable neonatal patients may benefit from initial conservative management, including prophylactic antibiotics and daily dressing changes to allow epithelialization of the omphalocele sac.

Cover of the heart needs to be accomplished without causing significant cardiac compression or distortion of the great vessels, resulting in decreased ventricular filling, reduced cardiac output, and death. It is well described that previous attempts to return the heart to a more “normal” anatomical position and close the defect with skin or myocutaneous flaps frequently produced fatal kinks of the great vessels. The materials to accomplish the closure of exposed body cavities include autologous tissues, cadaveric skin, and a range of alloplastic and biosynthetic materials. The ideal material should not interfere with the patient’s growth, should be pliable, soft, resistant to tearing, shrinkage and calcification, and not induce remodeling or scar tissue.

The PC treatment consists of corrective or palliative cardiovascular surgery, correction of ventral hernia and diaphragmatic defects and correction of associated anomalies. The surgical plan can be made as stepwise or a single operation depending on the patient clinical data, hemodynamic compatibility, thoracic cavity compatibility and the type of intracardiac defect. The best treatment strategy depends on the size of the abdominal wall defect, the associated heart anomalies, and the type of EC. The overall aims of surgical treatment are to provide soft tissue cover to the exposed heart and abdominal organs and reduce these into their respective cavities, palliation or repair intracardiac defects, and reconstruct the chest and abdominal walls. When planning the first stage of repair of EC, the following factor needs to be addressed: availability of tissue to cover the heart and abdominal viscera or lack thereof; exposure of open body cavities to the environment; disproportion in the size of the thoracic cavity and heart; and severity of the intracardiac lesion. Surgical treatment is complex, and various operative techniques often require a staged approach. The prognosis of this defect anomaly is poor, especially for thoracic and cervical types—the more extreme its position, the poorer the prognosis. Thoraco-abdominal type, however, has a better prognosis because of the slight abnormality of the gross anatomy of the heart and great vessel. The mortality rate is quite high, with a survival rate of less than 5%. Even the very mild or incomplete form of PC has only less than 40%. Atmospheric exposure to the heart makes it vulnerable to bacterial contamination that can lead to infection or even severe sepsis. Therefore this condition also contributes to patient morbidity and mortality.

**SUMMARY**

The coexistence of omphalocele and ectopia cordis characterizes the pathognomonic Pentalogy of Cantrell. Prenatal imaging studies are essential not only for prenatal counseling but also for adequate postnatal therapeutic planning. Prenatal diagnosis is important in guiding the surgical team to estimate the prognosis and reduce a postnatal management strategy. Early management can provide the basic care necessary to keep the patients until definitive management can be provided.

**CONFLICT OF INTEREST**

There is no potential conflict of interest relevant to this article reported.

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None.

**AUTHOR CONTRIBUTION**

All authors took part in this report, review, and manuscript.

**ETHICAL CONSIDERATION**

The patient’s parent has permitted their children’s data to include in this case report and to publish the article.

**REFERENCES**