# PENTALOGY OF CANTRELL WITH ECTOPIA CORDIS: A RARE CONGENITAL ABNORMALITY

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## ABSTRACT

The link of sternal fusion defects with many cardiac, diaphragmatic, and anterior body wall defects represents developmental field complex that includes the Pentalogy of Cantrell (PC) and Ectopia Cordis (EC). Attempts of surgical repair leads to survival of few numbers of patients and the main causes of death in these patients include tachycardia, bradycardia, hypotension, rupture of the diverticulum, and failure of heart. We report a diagnostic case of PC ina 12-hour baby girl in AFIC/NIHD Rawalpindi.



## INTRODUCTION

Ectopiacordis (EC) is one of the unique congenital abnormality manifested as the existence of a live beating heart outside the chest. Pentalogy of Cantrell is a very rare syndrome that links with varying proportions of midline wall defects and congenital cardiac deformities1. It is defined as a combination of five abnormalities that are: defect in a midline supra umbilical abdominal wall, a sternal defect, an anterior diaphragmatic defect, a diaphragmatic pericardial defect and a congenital intra cardiac defect. Frequency is rare including 1/100,000 births. Important attributecomprises of omphalocele linked with EC[2].

## CASE REPORT

We describe a case of 12-hour-old baby girl with 1.88 kg weight and 49 cm height who was born at term after uncomplicated pregnancy. She was tacypneic, mildly cyanosed with peripheral oxygen saturation around 80 percent and heart rate of 144 breaths/min. Physical examination shows complete EctopiaCordis (fig) linked to a large thoraco-abdominal wall. The heart protruded via complete sternal defect and was lying in inverse position outside the chest with apex pointing towards right shoulder. The defect of supra umbilical portion of abdominal wall was also complete and the omphalocele was covered by peritoneum. Transthoracic echocardiography revealed double-outlet right ventricle, moderate atrial septal defect, large subortic ventricular septal defect, pulmonary valvular as well as infundibular stenosis. Unfortunately, the newborn died in the first 24 hours from refractory septic shock.

## DISCUSSION

Pentalogy of Cantrell is congenital disorder which is a rare event and usuallycomprises combination of birth defects. It is characterized in 1958 by Cantrell, Haller and Ravich who described cardiac ectopia with other abnormalities of the mid-line and also includes supra-umbilical, lower sternum, pericardium and the anterior diaphragmaticdefects as Pentalogy of Cantrell[3]. Degree of severity varies andit potentially cause life threatening complications. Toyama suggested classificationinto class 1: with the presence of all five defects as complete syndrome; class 2: with the presence of four defects as probable

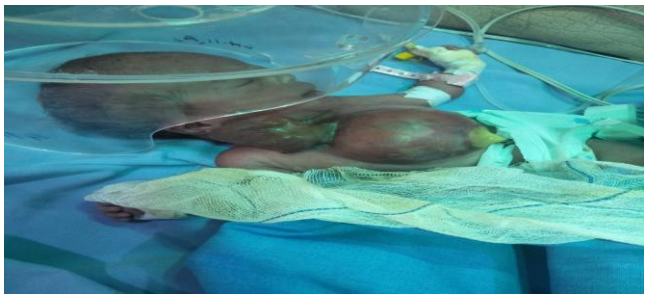

**Figure: Complete extra thoracic Ectopia Cordis and Omphalocele.**

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syndrome (including intra cardiac as well as ventral abdominal wall abnormalities); and class 3 as partial syndrome, having various combination of defects present[4]. Ectopia Cordis (EC) and omphalocele represents severe form of Pentalogy of Cantrell at birth. Omphalocele is an abdominal wall defect in which intestine protrude and which can be small, or it can be large, in which both intestines and abdominal organs protrude.

EC is a rare congenital defect in the union of the anterior chest wall resulting in location of the heart outside the chest. Its prevalenceestimated out to be 5.5–7.9 per million live births[5-6].

Embryonically, Ectopiacordisis a result of abnormal migration ofsplanchnic and somatic mesoderm and effects the development of the heart and the major vessels, with the premature rupture of the chorion or vitelline sac leading to a mid-line defect[7]. In complete EC the heart is completely present external to the thoracic cavity with or without pericardium. It presents as a new born emergency which is a direful condition and usually leads to deathdue to heart failure, sepsis, or hypoxemia. It's more commonly associated with intra cardiac defects when it is a part of Cantrell pentalogy. A broad range of associated cardiac abnormalities has been reported, the most common are ventricular septal defect, atrial septal defect, tetralogy of Fallot, pulmonary stenosis. Other cardiac deformities are patent ductusarteriosus, hypoplastic left heart syndrome, and univentricular heart with pulmonary stenosis[8].

The treatment options consistof corrective cardiovascular surgery and very few number of patients remain alive,prognosis seems to be bad in patientsespecially with the complete form of Pentalogy of Cantrell, total Ectopia Cordis, the main reasons of death including tachyarrhythmia, bradycardia, hypotension, rupture of the diverticulum, and heart failure[9].

## CONFLICT OF INTEREST

This study has no conflict of interest to be declared by any author

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