

## Case Report

# Pentalogy of Cantrell: case report and review of the literature

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## ABSTRACT

Pentalogy of Cantrell is a syndrome that consists of five anomalies: a midline, upper abdominal wall abnormality; lower sternal defect; anterior diaphragmatic defect; diaphragmatic pericardial defect, and congenital abnormalities of the heart. The pathogenesis of this condition is not fully known yet, associations are common with this condition and treatment is extremely challenging. Prognosis and outcome depends on the type of the pentalogy of Cantrell and associated cardiac anomalies. It can

be diagnosed reliably by antenatal ultrasound during the first trimester. We describe a Sudanese baby of undetermined sex who had the typical features of pentalogy of Cantrell and died shortly after birth.

### Key words:

Pentalogy of Cantrell; Anomaly; Diaphragmatic defect; Sudan

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## INTRODUCTION

In 1958, Cantrell et al [1] described a syndrome in which a ventral (anterior) diaphragmatic hernia occurred in association with an omphalocele. This syndrome, called the pentalogy of Cantrell (PC), consists of the following: A deficiency of the anterior diaphragm, a midline supraumbilical abdominal wall defect, a defect in the diaphragmatic pericardium, congenital intracardiac abnormalities, and a defect of the lower sternum. Few variants of this syndrome have been described [1,2].

Incidence of the PC varies from 5.5 to 7.9 per million live births [3]. The constellation of defects observed in PC is thought to result from the abnormalities in the differentiation of the intraembryonic mesoderm at approximately 14 to 18 days after conception. The diaphragmatic and pericardial defects result from abnormal development of the septum transversum whereas the sternum and abdominal wall defects are probably related to impaired migration of mesodermal structures [4].

Many other associations have been found with PC, which include the following: Amniotic band syndrome with limb anomalies, structural cardiac defects with pericardial effusion, exencephaly, cystic hygroma, infraumbilical defects with cloacal and bladder exstrophy, and bilateral inguinal hernias. Concurrent structural and/or chromosomal abnormalities may complicate up to 50% to 75% of cases presenting with omphaloceles and, thus, they are indicators for antenatal invasive testing [5,6]. If the maximum diameter of the omphalocele is greater than 1 cm in the first trimester or if it persists beyond 14 weeks of gestation, this is regarded as pathological [7,8].

We report a Sudanese baby with pentalogy of Cantrell in whom the diagnosis was made antenatally and confirmed immediately after birth. To the best of our knowledge, this is the first case report of pentalogy of Cantrell from Sudan.

## CASE REPORT

A 29-year-old Sudanese woman in her second pregnancy presented for the first time at 34 weeks gestation for antenatal care, she had a miscarriage before. Ultrasonography evaluation at 34 weeks revealed a viable foetus with major defects, absent anterior abdominal wall with herniation of the bowel and liver, absent lower third of the sternum with bulging heart, There was also a diaphragmatic hernia. These defects were consistent with pentalogy of Cantrell (Figure 1).



**Figure 1 – Antenatal ultrasound showing (A) protruded heart under the skin and (B) herniating viscera and liver.**

These findings were discussed extensively with the parents as well as the grave prognosis for the foetus. The parents decided to go for induction of labour. Labour was induced at 34 weeks + 6 days (Assisted -Forceps - vaginal delivery) , the outcome was a baby of undetermined sex, Apgar score was 4 at 1minute and 4 at 5minutes , Weight was 2 kg .The baby needed resuscitation in form of Ambu bagging and oxygen and was put on continuous nasal oxygen. Heart rate was 90 beats /minute, respiratory rate 60 breath /minute. The O2 saturation was ranging between 60-80%. Examination showed no obvious facial dysmorphic features, absent lower sternum with protruded heart,

large supraumbilical anterior abdominal wall defects with evisceration of oedematous bowel loops, liver and spleen. There was also imperforate anus, no external genitalia and left sided talipesequinovarus (Figures 2 and 3).

No investigations or further intervention was performed upon the request of the parents and the baby died at 45 minutes of age.



**Figure 2 - Imperforate anus and absent external genitalia (arrow).**



**Figure 3 – Evisceration of oedematous bowel loops, liver and spleen.**

## DISCUSSION

Pentalogy of Cantrell is a rare congenital anomaly, the pathogenesis of which has not been fully elucidated. Often the spectrum of the original pentalogy of Cantrell is not complete. Toyama [2] suggested the following classification of the pentalogy of Cantrell: class 1, definite diagnosis, with all five defects present; class 2, probable diagnosis, with four defects present, including intracardiac and ventral wall abnormalities; and class 3, incomplete expression, with various combinations of defects present, including a sternal abnormality. Our case possibly belongs to type 2.

Familial cases have been described, suggesting probable recessive inheritance [9]. There is one case report of PC with consanguineous parents [10]. In our case, there was no consanguinity.

With prenatal ultrasonography, the pentalogy of Cantrell usually can be diagnosed in the first trimester of pregnancy [11]. Our case was reliably diagnosed antenatally despite late presentation of the mother to the antenatal clinic at 34 weeks gestation. The visualization of the foetal anomalies can be enhanced by the use of prenatal magnetic resonance imaging (MRI) [12].

After birth, echocardiography is essential for diagnosis of associated cardiac anomalies. In our case, echocardiography was not done as the parents refused all investigations. Other features of the pentalogy of Cantrell and known associated anomalies can be diagnosed by conventional radiography or sonography. Nevertheless, small defects of the diaphragm and pericardium can be extremely difficult to diagnose accurately. In these patients and in cases of possible surgical intervention, MRI might be useful [13,14].

Associated anomalies are common in PC, our case had imperforate anus and left club foot which are frequently described [15,16]. Interestingly, our patient had no external genitalia and so far, this has not been

described as an associated anomaly.

Treatment of PC is challenging and outcome depends on the size of the abdominal wall defects, ectopia cordis and associated heart defects. The treatment consists of corrective or palliative cardiovascular surgery, correction of ventral hernia and diaphragmatic defects and correction of associated anomalies [17]. In 1972, Toyama reported a survival rate of 20% [2] whereas Ghidini reported a survival rate of 0% in a total of 17 patients [18]. Hornberger, in 1996, showed that mortality occurred at a rate of 50% in patients with ectopia cordis associated to a thoraco-abdominal defect [19].

In conclusion, PC is a very rare constellation of congenital anomalies that can be reliably diagnosed antenatally and prognosis depends on the type of PC and associated cardiac anomalies. The parents should be fully counselled regarding the prognosis of this condition.

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