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Case Report

Pentology of Cantrell: a rare case report with review of literature

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ABSTRACT

Pentology of Cantrell a rare congenital disorder of unknown etiology consists of pentad of: defect in the diaphragm, abdominal wall defect, deficiency of the pericardium and intracardiac defects. The hallmark diagnostic findings being ectopia cordis and omphalocele. The exact pathogenesis is not clear, and the prognosis of this lethal condition depends on the severity of anterior abdominal wall defect and associated cardiac anomaly. Antenatal ultrasonography is a reliable tool of diagnosis and allow for termination of pregnancy if done before 20weeks of pregnancy. First described by Cantrell in 1958, very few cases have been reported till now in the literature. Authors describe here a case of a neonate delivered at 32weeks of gestation weighing 2.3kg at their hospital with typical features of Cantrell's Pentology associated with a rare interesting finding of absent external genitalia and presence of lumbar meningocele.

Keywords: Ectopia cordis, Omphalocele, Pentology of Cantrell

INTRODUCTION

Pentology of Cantrell (thoraco abdominal syndrome) first described by Cantrell et al in 1958, is a rare genetic disorder of unknown etiology.¹ The pentad consists of:

- Ectopia cordis and intracardiac anomalies;
- lower sternal defect;
- midline supraumbilical thoraco-abdominal wall defect;
- anterior diaphragmatic defect; and
- defect of diaphragmatic part of pericardium that results in relation between pericardial cavity and peritoneum.

Concurrent structural &/or chromosomal defects complicate upto 50-70% of cases. The exact pathogenesis is not known, thought to result from defect in the embryogenesis (6-7th week of pregnancy) due to abnormal migration of sternal anlagen and myotomes. The

incidence is very rare, ranging from 5.5 -7.9 million live births.²

The prognosis of this condition is very poor and depends on the severity of anterior abdominal wall defects and associated intracardiac anomaly.

CASE REPORT

A baby delivered prematurely at the labor room at ESIC Hospital at 32 weeks of pregnancy, weighing 2.3kg with very poor APGAR score. The history of mother G2P1L1 revealed no regular antenatal checkups, no intake of folic acid or iron supplements, no anomaly scan at 18-20weeks, no history of exposure to teratogenic drugs/radiation. Previous normal vaginal delivery of female baby, alive and healthy. There was no history of significant medical illness in either of the parents or other family members. On further history she revealed of third degree consanguineous marriage. On general and

abdominal examination no abnormality was detected. In view of poor prognosis associated with lethal abnormality, the parents consented for “do not resuscitate”. The patient and husband consented for autopsy but refused any further genetic testing.

On gross examination of the fetus there was ectopia cordis, large anterior abdominal wall defect with protrusion of liver and loops of bowel (Figure 1), absent external genitalia & absent anal opening (Figure 2), meningocele at the lumbar region and contractures of limb.



Figure 1: Fetus showing ectopia cordis, with evisceration of liver, loops of intestine through anterior abdominal wall defect, and contractures of limbs.



Figure 2: Absent external genitalia imperforate anus.



Figure 3: Dissection of heart showing pulmonary stenosis and ductus arteriosus.

Fetal autopsy revealed absence of the distal third sternum, absence of pericardium, absence of the diaphragm, and associated cardiac anomalies viz., patent ductus arteriosus, pulmonary stenosis (Figure 3) with left ventricular hypertrophy but no septal defects.

DISCUSSION

Cantrell offered a developmental failure in lateral mesoderm during day 14-18 after conception. The diaphragmatic and pericardial defects result from abnormal development of the septum transversum whereas the sternum and abdominal wall defects are probable due to impaired migration of mesodermal structures.¹ Most cases are sporadic, although found association with familial inheritance. There is one case report of PC with consanguineous marriage, and present case also reports consanguinity of third degree.³

Toyama suggested following classification: class 1, with all five defects present; class 2, with four defects present, including intracardiac and ventral wall abnormalities; and class 3, incomplete expression, with various combinations of defects present.⁴ The present case fits into class 1 or complete variety.

Associated anomalies are common in PC, the present case had imperforate anus, contractures of upper and lower limbs which have been described in other studies.^{5,6} In addition the present case had a very interesting finding of absent external genitalia which has been described in only one case before till now.⁵ There have been reports of association with Trisomy 18, 13 and Turners syndrome, chromosomal analysis is recommended. Fetal Echo and MRI may be useful to look for associated structures defects and also for prognostication and planning the further management.⁷

Prognosis of PC is very poor, and survival depends on severity of abdominal wall defects and associated intracardiac anomaly. Toyama, in 1972, reported a survival rate of 20%⁴, whereas Ghidini reported a dismal survival of 0% in total of 17 patients.⁸ Norma et al., reported outcome of 22 patients surgically treated for PC where babies without extracardiac defects had favourable outcome.⁹ In 1996, Hornberger reported mortality of 50% in patients with ectopia cordis associated to a thoraco-abdominal defect.¹⁰ In view of poor prognosis and few survival rate after corrective surgery, termination of pregnancy may be considered, if diagnosed before viability by ultrasound.

CONCLUSION

The present case had all the features of PC fitting in the class I by Toyama classification, with rare interesting finding of imperforate anus and absent external genitalia reported in only one case till now. With prenatal ultrasonography, PC can be diagnosed easily in the second trimester before 20 weeks. The purpose of this

study emphasizes the importance of routine anomaly scan in second trimester, thus allowing for termination of pregnancy.

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