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Case Report

Pentalogy of cantrell with single umbilical artery: a rare fetal anomaly

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ABSTRACT

Pentalogy of cantrell (PC) is a rare syndrome having both specific ventral and dorsal midline defect, and rarely found to be associated with other anomalies. Most cases occur randomly for no apparent reason (sporadically). We report a 20 years old primigravida referred at 17 weeks 4 days of gestational age, for a fetal anomaly scan having an anterior abdominal wall defect in fetus. Ultrasound done, showed single live fetus gestational age of 18 weeks, with fetal heart, great vessels, stomach, and intestinal loops were found to be lying outside thoracic cavity. Fetal autopsy following termination of the pregnancy confirmed the presence of the malformations with single umbilical artery. Occurrence of both ventral and dorsal midline defects suggests that the timing of fetus insult would be between 14 to 18 days after conception when the splanchnic mesoderm (myocardium) and somatic mesoderm (sternum, abdominal wall, diaphragm and pericardium) is dividing.

Keywords: Ectopic cordis, Pentalogy of cantrell, Single umbilical artery

INTRODUCTION

Pentalogy of cantrell (PC) syndrome first described by Cantrell and colleagues in 1958 consisting five anomalies, a lower sternum defect, an anterior diaphragmatic defect, a diaphragmatic pericardial defect, an abdominal wall defect, and intracardiac abnormalities.[1] It has a rare frequency of about 1/100,000 births.[2] Study report here an antenatally diagnosed case of pentalogy of cantrell associated with single umbilical artery.

CASE REPORT

A 20 year old primigravida at 17 weeks 4 days gestational age referred to our antenatal OPD, with a fetal anomaly scan (an ultrasound done outside had shown some evidence of an anterior abdominal wall defect). The patient's marriage was non-consanguineous, and she had no significant medical and surgical illness or history of any genetic disease, recurrent abortion or medication (except folic acid and iron).

On examination, she had normal general physical examination and systems examination. Ultrasound done at our clinic centre showed single live fetus of 18 weeks, with fetal heart, great vessels, stomach, and intestinal loops were found to be lying outside thoracic cavity and single umbilical artery in cord. Facial appearance and extremities appeared to be normal.

The couples were counselled, opted for pregnancy termination. Amniotic fluid examination for karyotyping reported normal (46 XX) and mother's TORCH profiles was reported negative. Fetal autopsy showed absent anterior diaphragm, omphalocoele, cardiac ectopia, absent of terminal portion of sternum and single umbilical artery on cord examination. Foetogram showed normal extremities and skull cranium.

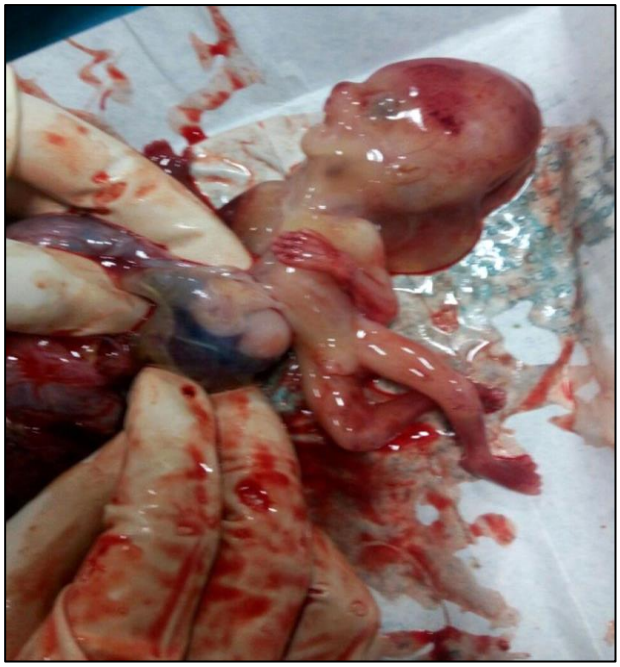

Figure 1: Pentalogy of cantrell: fetus with omphalocoele SAC.

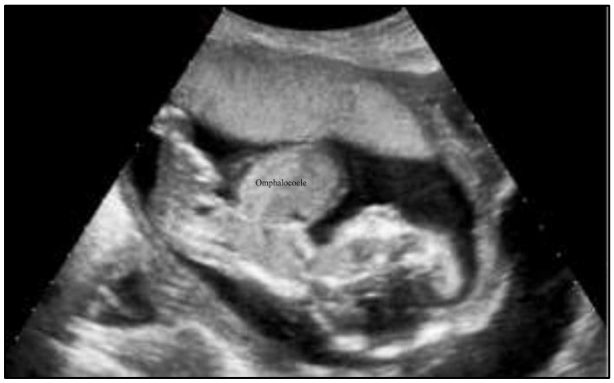

Figure 2: Pentalogy of cantrell: omphalocoele.

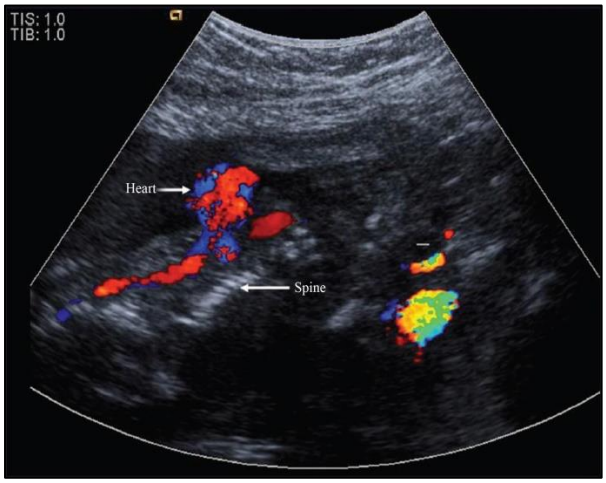

Figure 3: Fetal ultrasounds: ectopia cordis.

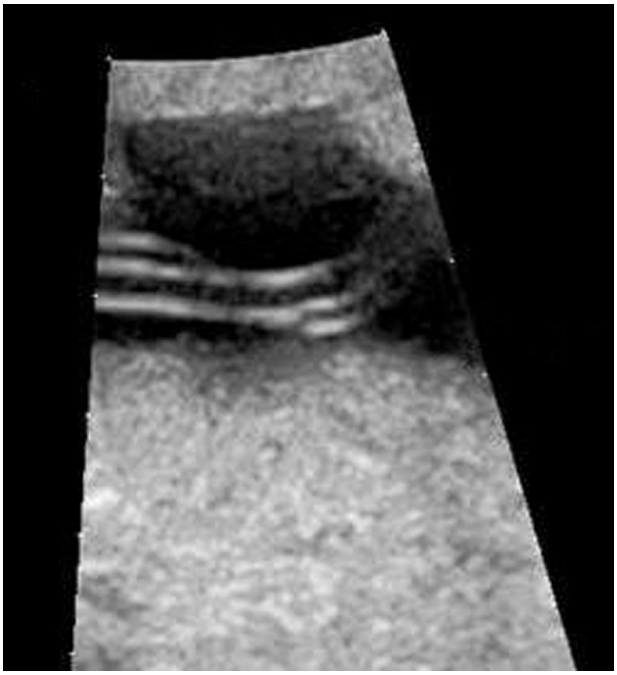

Figure 4: Pentalogy of cantrell: single umbilical artery.

DISCUSSION

Pentalogy of cantrell is rare congenital anomaly first described by cantrell in 1958.[1] Complete PC is defined when all five criteria described by cantrell are there but incomplete form having three or four described criteria's. Range of each defect is variable, as sternal defect can range from absence of xiphoid to shorting, cleaving, or absent sternum. The abdominal defect ranges from wide rectus muscle diastasis to a large omphalocoele.[3]

Abdominal wall, sternum, pericardium, and diaphragm arise from somatic mesoderm, while myocardium arises from splanchnic mesoderm. The proposed embryogenesis postulates a failure of the lateral mesoderm folds to migrate to the midline, results in sternal and abdominal defects, and septum transversum failure result in anterior diaphragm and pericardium defects. An insult occurring prior to differentiation of the mesoderm into these two layers could produce defect in all affected structure.[1] These insults could be varied such as mechanical disruption via amniotic bands, single gene mutations, chromosomal abnormalities (particularly trisomy 18) and disruptive vascular events.[4]

Our case also had an additional defect in form of single umbilical artery along with ventral and dorsal defects.

Imaging should be performed to rule out associated anomalies. Foetal echocardiography is performed to evaluate and diagnose any cardiac abnormalities. Foetal MRI may be beneficial in selected cases.[5] Overall the prognosis appears dismal but may be related to the extent of the defect. Toyama demonsted a survival rate of 20%

in this order including its variants and incomplete syndromes.[6] The complete pentalogy has a poorer outcome, and the survival rate was only 8.5% in the report of Fernandez et al.[7] Termination of pregnancy is considered if ultrasound diagnosis is made before viability in view of poor prognosis. There is no data available till date that showing improved or changed outcome with caesarean delivery. Surgical intervention in form of repairing sternal, diaphragmatic, pericardial defect and abdominal defect can be done at same time. Case report is submitted for three reasons; one is that it is a very rare syndrome, second for highlight the second trimester anomaly scan's importance and third one as it has rare association with single umbilical artery.

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