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Case Report

Incomplete pentalogy of Cantrell: a case report

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ABSTRACT

Pentalogy of Cantrell (PC) is a rare congenital anomaly characterized by a defect in the lower sternum, anterior diaphragm, and anterior abdominal wall; ectopia cordis; and congenital heart disease. Authors report a case of male foetus terminated at 20 weeks of gestation with an Incomplete (class 3) pentalogy of Cantrell presenting with gastroschisis, ectopia cordis and absence of lower sterna. Prognosis of pentalogy of Cantrell depends on severity of intra and extra cardiac defects, pulmonary hypoplasia, extent of abdominal wall defect, cerebral anomalies and diaphragmatic herniation. Full pentalogy of Cantrell is a severe and rare syndrome, but incomplete forms with combination of two or three defects are reported frequently similar to present case report. Early diagnosis through obstetric ultrasound would help in improved informed clinical decision making on the part of the obstetrician and family.

Keywords: Abdominal wall defect, Congenital, Ectopia cordis, Gastroschisis, Incomplete, Pentalogy of Cantrell (PC)

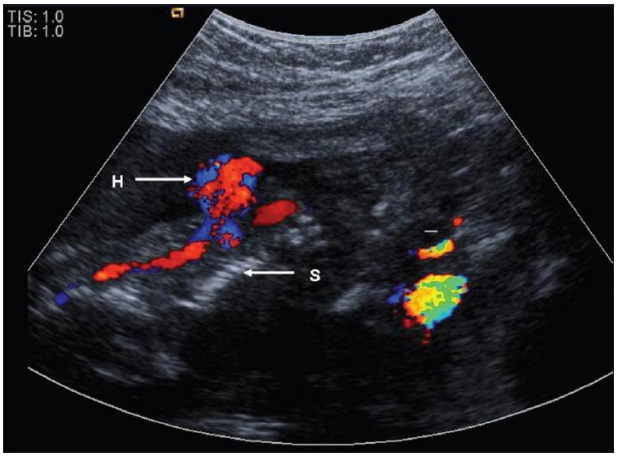
INTRODUCTION

Cantrell first described the full spectrum of PC.[1]The Pentalogy of Cantrell, PC is an extremely rare and usually lethal anomaly. The full spectrum consists of five anomalies: A deficiency of the anterior diaphragm, a midline supraumbilical abdominal wall defect, a defect in the diaphragmatic pericardium, ectopia cordis various congenital intracardiac abnormalities, and a defect of the lower sternum.[1] Authors report a case of incomplete Pentalogy of Cantrell. Most cases are diagnosed early in the second trimester, when ectopia cordis associated with gastroschisis or omphalocele is observed.[2] The first description of PC was made by Cantrell et al in 1958, who reported 5 cases with this anomaly.[1] Up to now, there are some case reports that have been classified as full spectrum, which have all of the five defects and incomplete forms with lesser defects of the pentalogy of Cantrell. An associated intracardiac anomaly is the rule, such as septal defects, Ebstein's anomaly, Fallot's tetralogy or single atrium. In 75% of the cases pericardium may be absent.

CASE REPORT

A 27-year-old G2 IUD1, booked case on regular antenatal care at Travancore Medical College hospital had dating scan corresponding to POG and low risk NT scan. Her medical history w unremarkable. Parents were non-consanguineous on routine anomaly scan at 20 weeks gestation, foetus in breech presentation with growth corresponding to POG showed a large abdominal wall and partially chest wall defect through which abdominal organs and cardia protruded out with no membranous covering with a diagnosis of gastroschisis.

There was spinal deformity (kyphoscoliosis) with pelvic deformity and rotation of lower limbs.4 D Ultrasonogram and MRI confirmed the above findings. No cardiac malformation was noted (Figure 1 and 2).



H-heart, S-spine.

Figure 1: Fetal ultrasound scan showing the ectopia cordis.

The parents were counselled regarding prognosis of the foetus and offered termination of pregnancy. With informed consent, oral mifepristone 200mg followed by one dose of 400µg vaginal misoprostol was given.

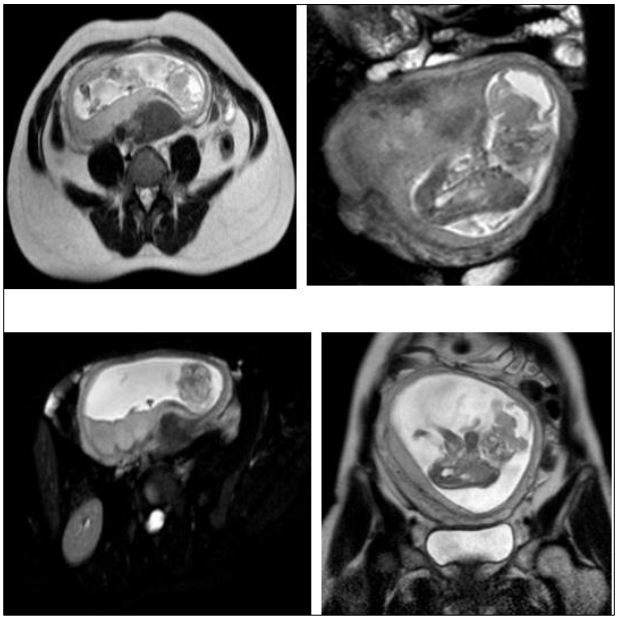


Figure 2: MRI showing features suggestive of gastroschisis, ectopia cordis.

She expelled a foetus weighing 200grams showing ectopia cordis, lower sternal defect, large supraumbilical anterior abdominal wall defect with liver, spleen and loops of small intestine and colon with tinea protruding outside characteristic of incomplete PC along with kyphoscoliosis spinal deformity and club foot (Figure 3 and 4). The thoraco-abdominal wall defect was extending from the umbilicus to the upper third of the body of the sternum. The liver was enlarged with a prominent caudate lobe.

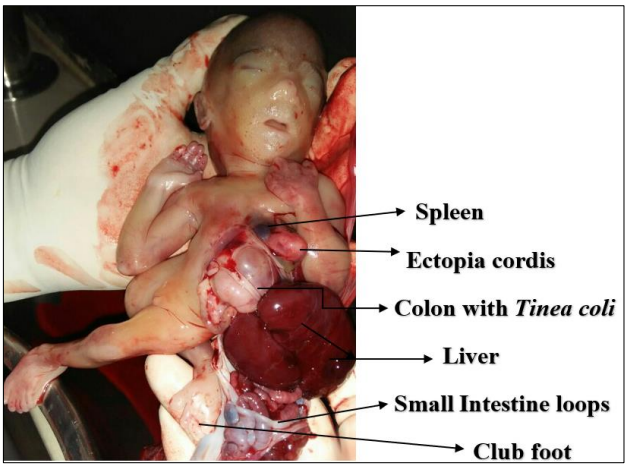


Figure 3: The baby at 20 weeks with the incomplete pentalogy of Cantrell having lower sternal defect with thoracoabdominal ectopia cordis, entire liver, spleen, gastroschisis with intestinal loops (both small and large) and kyphoscoliosis with pelvic deformity with rotation of lower limb and club foot.

Consent for autopsy was denied by the parents and hence not done. The external genitalia showed a normal male phenotype.

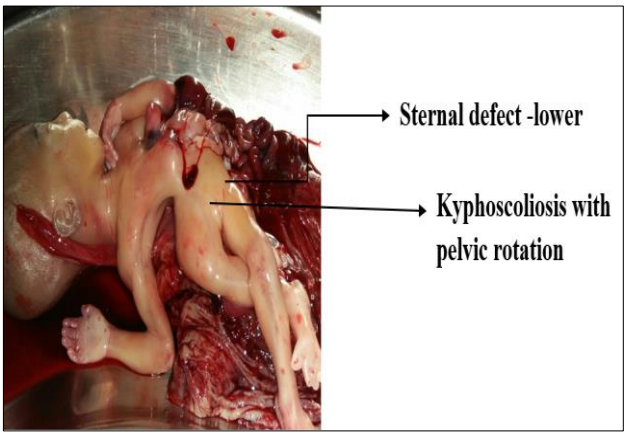


Figure 4: The baby at 20 weeks with the incomplete pentalogy of Cantrell having kyphoscoliosis with pelvic deformity and rotation of lower limb; club foot is also noted.

DISCUSSION

Cantrell's pentalogy consists of: midline supra-umbilical abdominal wall defect, defect of the lower sternum, deficiency of the anterior diaphragm, defect in diaphragmatic pericardium, congenital intracardiac defects.

Classification

Toyama suggested the following classification for Pentalogy of Cantrell[3]:

- Class I: definite diagnosis with five defects present.
- Class II: probable diagnosis with four defects (includes intracardiac and abdominal wall defects).
- Class III: incomplete expression with combination of defects (but with the presence of sternal abnormality).

Present case fits into Class III PC with midline supraumbilical abdominal wall effect, defect of lower sternum and thoracoabdominal ectopia cordis.

Embryology

According to Cantrell et al, PC is due to failure of development of ventral mesoderm at about 14 to 18 days of embryonic life Complete or incomplete failure of midline fusion results in a range of related malformations from isolated ectopia cordis to complete ventral evisceration.

On the basis of embryonic development this syndrome may be classified into two groups[4]:

- Due to mesodermal defect which includes defects in diaphragm, pericardium and intracardiac lesions.
- Due to failure of migration of paired structures which includes sterna and abdominal wall defects.

Because of various phenotypes of abdominal wall defect in Pentalogy of Cantrell, multiple factors are said to be responsible, including mechanical teratogens, major gene mutations, chromosomal abnormalities such as trisomy 13 and trisomy 18 and disrupted vessels defects. Mutations of TAS gene which mapped at Xq25-q26.1 area, is mentioned to have a role in fusion of sternum, multiple cardiac, diaphragmatic, anterior abdominal defects and additional abnormalities.[5] Perixoto and Filho et al, mentioned club foot in few cases. Authors also had club foot in present foetus.[6]

Incidence

Complete pentalogy of Cantrell (PC) is a rare condition with an incidence of 1 in 100000 pregnancies.[1]: PC1 consists of: Ectopia cordis and intracardiac anomalies, lower sternal defect, midline supraumbilical thoraco-abdominal wall defect, anterior diaphragmatic defect, defect of diaphragmatic part of pericardium that results in relation between pericardial cavity and peritoneum.

Most cases are sporadic, and no recurrences have been reported. There is male dominance with a male to female ratio of 2.7:1 as in ours. An incidence of ectopia cordis was reported to be 5.5 to 7.9 per 1 million live births by Chandran S. et al, Alphonso N et al. cited the same incidence but limited it to thoracic ectopia cordis.[2,7]

Ectopia cordis occurs when the heart is displaced outside the chest wall.[8] The displacement of the heart can be cervical, cervico-thoracic, thoracic, thoraco-abdominal, or abdominal. The most common types are thoraco-abdominal and abdominal an associated intracardiac 1anomaly is the rule, such as septal defects, (VSD) in 100% of cases, ASD in 50% of cases, Ebstein's anomaly, Fallot's tetralogy in 33% of cases or ventricular diverticulum in 20% of PC. In 75% of the cases pericardium may be absent. Extracardiac abnormalities.[9] in PC include craniofacial anomalies such as cleft lip and/or palate; central nervous system.[10] anomalies such as hydrocephalus; skeletal malformations such as clubfoot, absence of tibia or radius and abdominal abnormalities such as polysplenia and gallbladder agenesis. sternal fusion defect is a rare malformation and an inferior type is seen in pentalogy of Cantrell. Sternal defects include bifid sternum (26%), absent xiphoid (10%) and absent lower 2/3 of sternum (9%). Anterior abdominal wall defect is mostly an omphalocele (63%).[11] A ventral retrosternal defect of the diaphragm occurs in 91% of the cases. Omphalocele, cardiac septal defects, absent pericardium, cleft lip and defects in the sternum and diaphragm were noted in the reported case as well.

Diagnosis/imaging studies

Pentalogy of Cantrell can be suspected by antenatal ultrasound. It has been suggested that magnetic resonance imaging and prenatal fetal echocardiography provide optimal assessment of fetuses with PC. Using 2D ultrasound in the first trimester, and the adjunctive use of the 3D ultrasound may help to enhance the visualization of the fetal anomalies in different orthogonal planes, even in unfavorable fetal positions. Intrauterine diagnosis of abdominal wall defects before 12 weeks is not possible as herniation of bowel out of abdomen is a normal event in foetal development at that time. After 12 weeks, the differential diagnoses include omphalocele, pentalogy of Cantrell and gastroschisis.[12] If diagnosis is made by ultrasound, screening for chromosomal abnormalities is recommended. Following prenatal diagnosis, termination may be offered in severe cases especially with abnormal karyotype.

Prognosis

Ghidini et al, in his analysis on ten cases of complete PC which were diagnosed antenatally by ultrasound observed poor prognosis.[13] Omphalocele, cardiac septal defects, absent pericardium, cleft lip and defects in the sternum and diaphragm were noted in the reported case as well Prognosis of pentalogy of Cantrell depends on severity of intra and extra cardiac defects, pulmonary hypoplasia, extent of abdominal wall defect, cerebral anomalies and diaphragmatic herniation.

Treatment

The treatment strategy and prognosis depend on the size of the abdominal wall defect, the type of EC, and the associated anomalies. A multidisciplinary team should follow-up milder forms in order to determine the best

time for delivery. After delivery, repair of the omphalocele should not be delayed. Repair of the sternal, diaphragmatic, and pericardial defects can be attempted at the same time.[14] Surgical correction is often difficult due to the hypoplasia of the thoraco-abdominal cavity and also inability to enclose the ectopic heart The mean survival rate without any interventional surgery is about 36 hours.[15] Studies showed that even with care monitoring in professional centers and multiple corrective surgeries, they had high morbidity and mortality rate and long-time prognosis is poor.[16] The limitations in some of them could be financial support and technical expertise to deal with.[17]

CONCLUSION

Diagnosis of pentalogy of Cantrell cannot be made unless radiologists look into when multiple congenital anomalies are diagnosed during fatal morphology ultrasonogram. Awareness regarding this rare diagnosis among obstetricians is a must and parents should be appropriately counselled regarding the prognosis before decision about termination of pregnancy in previable foetuses.

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