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RESEARCH ARTICLE

INCOMPLETE PENTOLOGY OF CANTRELL: ULTRASOUND FINDINGS AND LITERATURE REVIEW OF THIS RARE CONGENITAL ANOMALY

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ABSTRACT

Pentalogy of Cantrell is a rare syndrome which was first described in 1958 by James R Cantrell. As this syndrome is rare, it becomes more important for the radiologist to know about wide range of anomalies that can be easily detected by ultrasonography leading to early diagnosis and appropriate management. We present here a case of 23 year old primigravida whose ultrasound abdomen was done for fetal well being which showed anterior thoracoabdominal wall defect in fetus with herniation of the heart, liver and gut loops into the amniotic cavity.

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INTRODUCTION

Pentalogy of Cantrell (also known as Cantrell's pentalogy or the Cantrell syndrome) is a rare syndrome which was first described in 1958 by James R Cantrell [1]. It includes 5 congenital anomalies: intracardiac defect, sternal cleft, pericardial defect, diaphragmatic defect, and epigastric omphalocele [1,2]. This syndrome has an estimated incidence of 1 per 65,000 live births [3]. As this syndrome is rare, it becomes more important for the radiologist to know about wide range of anomalies in this syndrome that can be easily detected by ultrasonography leading to early diagnosis and appropriate management. During intrauterine life ultrasonography (USG), MRI can be safely used for the detection of the congenital anomalies in the fetus. Ultrasound is safe, non ionizing, cost effective, easily available modality for diagnosis of intrauterine fetal anomalies like POC.

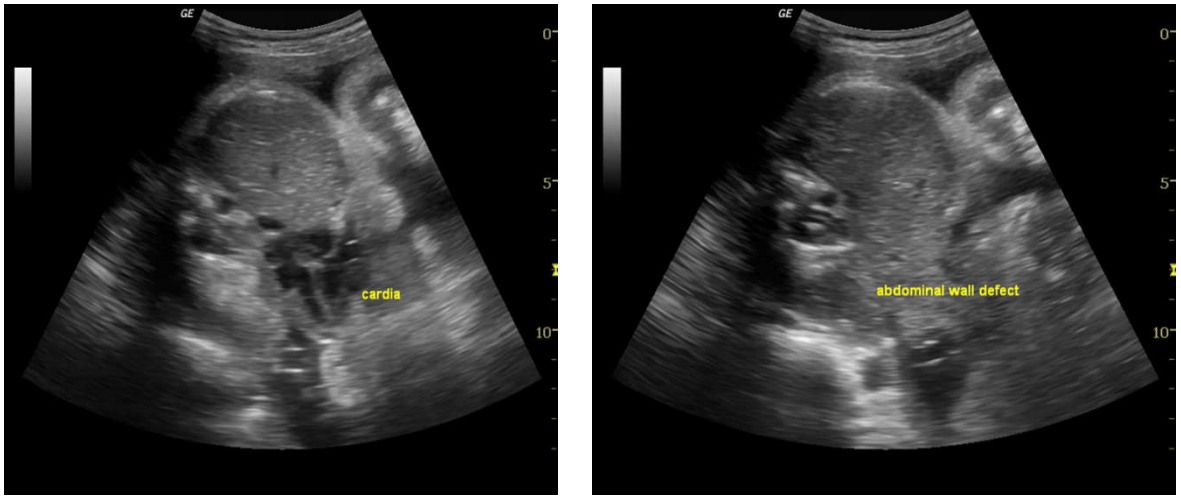
Case Description

23 year old primigravida with nine months amenorrhoea presented with pain abdomen and was referred to radiology department for antenatal ultrasound to know fetal well being, expected fetal weight, amniotic fluid index and estimation of gestational age. She had no history of bleeding, leakage per vaginum, medicine intake during pregnancy, cardiac or other congenital anomalies in her family.

Transabdominal sonography using GE machine with a 3.5 MHz convex probe was done and it showed single live intrauterine fetus with gestational age of to 36 weeks, expected fetal weight was 2058 grams. The location of placenta was posterior and not low lying. The amniotic fluid was grossly normal. There was presence of anterior lower thoracic upper abdominal wall defect with herniation of the heart, liver and gut loops into the amniotic cavity [Figure1]. The herniated liver, both small and large bowel loops were seen floating in the amniotic cavity [Figure 2]. Umbilical cord was attached to the abdominal wall just below the defect and demonstrated two arteries and one vein. Head and spine appeared normal. Stomach bubble was visualised. Both the kidneys, urinary bladder were normally visualised. No pleural effusion, pericardial effusion, fetal ascites was seen. On fetal echocardiography, fetal heart appeared structurally and functionally normal.

DISCUSSION

Pentalogy of Cantrell (POC) is rare congenital syndrome, first reported by James R Cantrell in 1958 [1], with incidence of 5.5 per million births and a 2:1 male to female predominance [4,5]. This syndrome consists of a midline anterior ventral wall defect, a defect of anterior diaphragm, a cleft distal sternum, a defect of apical pericardium with communication into the peritoneum, and an intracardiac defect [6]. Every case of Pentalogy of Cantrell don't present with all 5 classical findings [1,2].



(a) (b)

Figure 1(a,b). Gray scale transabdominal ultrasound images of the fetus in a transverse section showing anterior thoracoabdominal wall defect with herniation of the heart and abdominal contents into the amniotic cavity.

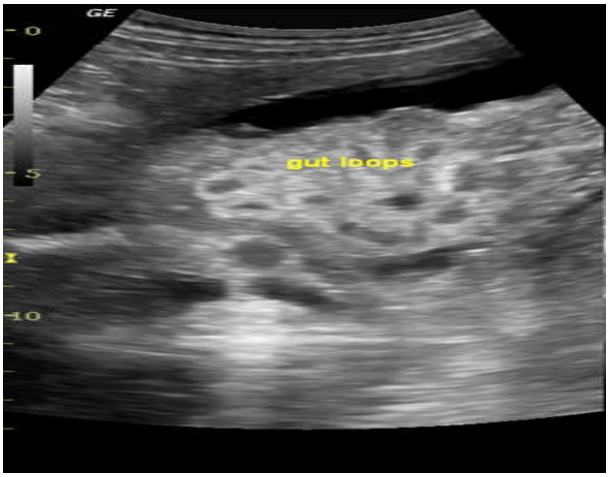


Figure 2. Gray scale transabdominal ultrasound images showing free floating herniated gut loops in the amniotic cavity

Toyama in 1972, classified Cantrell's pentalogy in three categories based on the number of finding: class I presents with all 5 defects and is a definite diagnosis; class II presents with 4 of the 5 defects and is a probable diagnosis; and class III presents with varying combinations of defects and is considered an incomplete expression. [7]. According to this, patient in our case can be categorised into class III. During the embryonic life between 14 to 18 days of gestation, there occurs developmental failure in the primordial lateral plate mesoderm leading to arrest of the thoracoabdominal wall closure [4,8]. Maximum cases of POC are sporadic, while few cases are also associated with trisomy 18 and X-linked inheritance[9]. Various anomalies associated with pentalogy of Cantrell are cleft lip, cleft palate, encephalocele, hydrocephalus and craniorachischisis, lung hypoplasia, adrenal hypoplasia, gallbladder agenesis, single renal agenesis, polysplenia, malrotation of the colon, herniation of bowel into pericardium, bladder exstrophy, undescended testes, bilateral inguinal hernia, club foot, absence of tibia, radius and hypodactyly[5]. This condition is classified into 4 types based on the position of the heart. In cervical ectopic cardia (3% of cases of ectopia cordis), the heart is displaced superiorly into the area of the neck. In the thoracic variety of ectopia cordis (60% of cases of ectopia cordis), the heart protrudes anteriorly through a sternal defect.

In the thoraco-abdominal variety of ectopia cordis (7% of cases of ectopia cordis), the heart is displaced outside the chest through a defect in the lower sternum in association with diaphragmatic and ventral abdominal wall defects. In the abdominal variety of ectopia cordis (30% of cases of ectopia cordis), the heart is displaced inferiorly into the abdomen through a defect in the diaphragm [10]. The main differential diagnosis is the omphalocele–exstrophy–imperforate anus–spinal defects complex, which is characterized by a combination of omphalocele, exstrophy of the bladder, an imperforate anus and spinal defects[5].

Conclusion

Radiology plays an important role in early diagnosis of pentology of Cantrell and associated anomalies during the intrauterine life. Ultrasound is safe, non ionizing, cost effective, easily available imaging modality useful in the diagnosis of POC. Fetal cardiac doppler helps in better evaluation of cardiac anomalies. MRI can also be used for evaluation of congenital fetal anomalies in cases where fetus can't be optimally evaluated by ultrasonography due to conditions like improper fetal position and oligohydramnios. These modalities help in early diagnosis of the anomalies so that timely intervention can be done.

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