

MRI Findings in Pentalogy of Cantrell with Ultrasound Correlation: Addis Ababa, Ethiopia

Leul Adane^{1*} Alemayehu Bedane², Ashenafi Abera², Fitehanegest Tefera² and Satyasai Panda³

¹Saint Paul Hospital Millennium Medical College, Department of Radiology, Consultant Radiologist, Addis Ababa, Ethiopia.

²Saint Paul Hospital Millennium Medical College, Department of Radiology, Consultant Radiologist, Ethiopia.

³Saint Paul Hospital Millennium Medical College, Department of Forensic Medicine & Toxicology, Professor Oreignsic Pathologist, Ethiopia.

*Correspondence:

Leul Adane Chemed, MD, Consultant radiologist, Department of radiology, Saint Paul Hospital Millennium Medical College, Addis Ababa, Ethiopia, Tel: +251912051411, E-mail: leuladane2@gmail.com.

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ABSTRACT

Pentalogy of Cantrell consists of an extensive defect of the thoraco-abdominal wall, which has nearly always a lethal prognosis. The defect is characterized by the association of five anomalies: omphalocele, cardiac ectopia, absence of the distal portion of the sternum, absence of the anterior diaphragm and absence of the parietal diaphragmatic pericardium. It has a rare frequency of about 5.5 per 1,000,000 live births. There is a common association with intra cardiac anomalies such as ventricular septum defect, tetralogy of Fallot, and transposition of great vessels. The pathogenesis remains unclear. Here we present an imaging findings with antenatal Two Dimensional (2D) and Three Dimensional (3D) ultrasound and fetal Magnetic Resonance Imaging (MRI) in a 20 weeks of gestation with a multiple anomalies, based on which the diagnosis of complete Pentalogy of Cantrell was given with a brief literature. Post mortem radiography, 3D Computed Tomography (CT) and clinical autopsy were performed additionally to enhance the visualization of fetal anomalies and to confirm the diagnosis.

Extensive imaging of cardiac, thoracic and abdominal malformations by ultrasound and MRI is complementary for a clear diagnosis and counseling of the patient.

Keywords

Pentalogy of Cantrell, Omphalocele, Gastrichisis, Ectopia cordis.

Introduction

The pentalogy of Cantrell was first described in 1958. The whole mark of this syndrome is an omphalocele associated with ectopia cordis. The full spectrum consists of five anomalies. A deficiency of the anterior diaphragm, a midline supra umbilical abdominal wall defect, a defect in the diaphragmatic pericardium, various congenital intracardiac abnormalities and a defect of lower sternum. Only a few cases with a full spectrum of the pentalogy have been described. We reviewed a literature to find the best imaging approaches in prenatal diagnosis, prognostic factors, and the best multidisciplinary approach in prenatal counseling and management in patients with pentalogy of Cantrell.

Case Report

Mrs. S came to our hospital in August 2017 with a complaint of back pain after she had sustained a trauma after an RTA. She is primigravida mother who claimed to be amenorrhic for 5 months, had no ANC follow up. She didn't feel fetal quickening. No history of vaginal bleeding or abdominal or pelvic pain. She has no history of any Alcohol intake or Smoking. Has no history of drug intake or exposure to any teratogenic agents, abdomino-pelvic examination revealed a twenty weeks pregnant uterus.

Her routine examinations were all in the normal range. Trans abdominal obstetrics ultrasound showed a single intrauterine alive male fetus with a corresponding GA (using BPD, HC and FL) of 19 wks. Amniotic fluid index and placenta were normal.



Figure 1: A small abdominal cavity with Evisceration of liver, stomach, spleen, and intestines are seen through an upper midline anterior abdominal wall defect without any surrounding membrane.

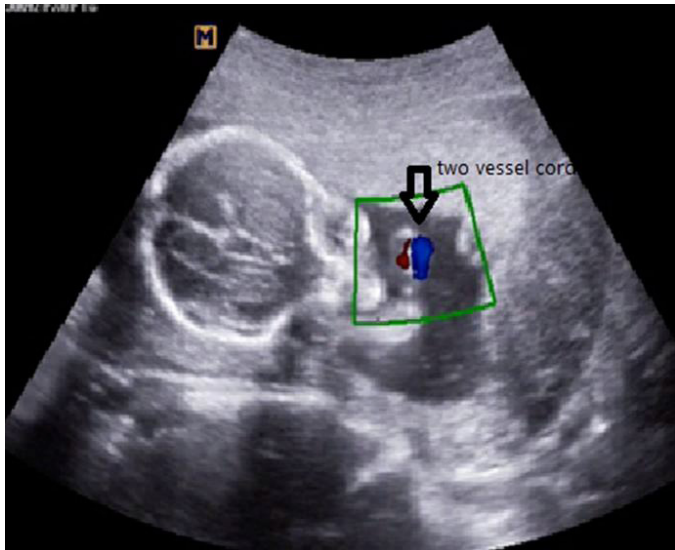


Figure 2: A two vessels umbilical cord was seen passing lateral and to the left of the defect and inserted on to the abdominal wall.

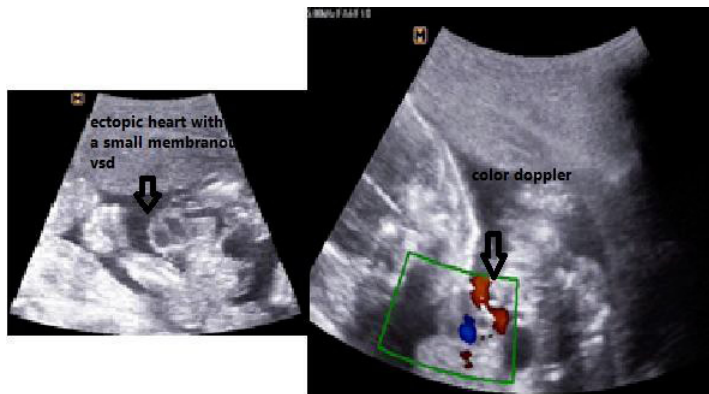
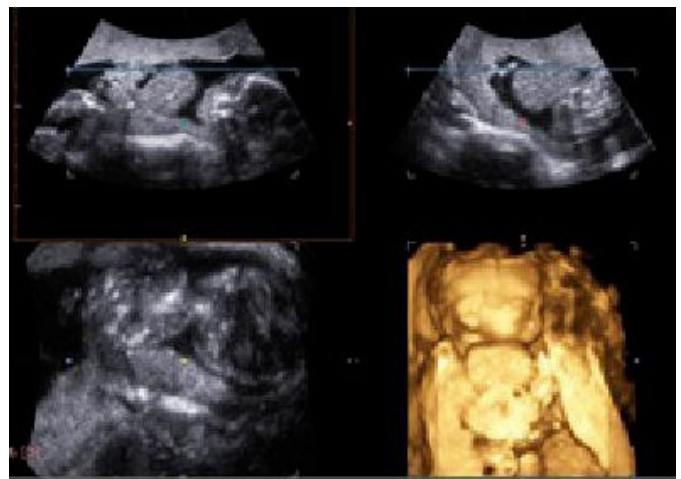


Figure 3: There is a small thoracic cavity with a hypoplastic lung and also a lower anterior chest wall defect heart lying outside the thoracic cavity. There is a four chamber heart, there is a small a membranous VSD.



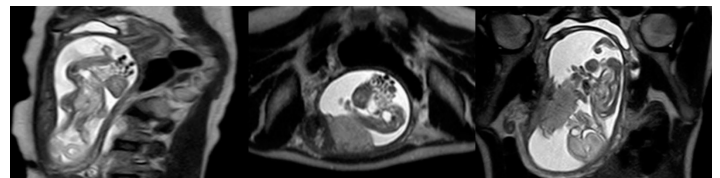
Figure 4: There is a clubbing deformity of the left foot.



3D ultrasound evisceration of the thoraco abdominal organs

Figure 5: A midline defect and evisceration of the thoracoabdominal organs seen on 3D ultrasound.

Obstetric MRI confirmed the ultrasound findings with greater precision of anatomical structures, also can observe absence of the hemi diaphragms and a continuous thoraco-abdominal cavities (Figures 6-8).



Figures 6,7,8: Coronal, axial and sagittal T2 BFFE, SSTSE fetal MRI; shows a continuous thoracoabdominal cavity with heart, liver, bowel loops protruding through the lower thoracic and upper anterior abdominal wall defects and freely floating in the amniotic fluid.

We informed the mother of the findings, after which she received medical counseling by obstetric and gynecology unit of the hospital for a therapeutic termination due to a fetal malformations with a very poor prognosis for the baby, and agreed. One week

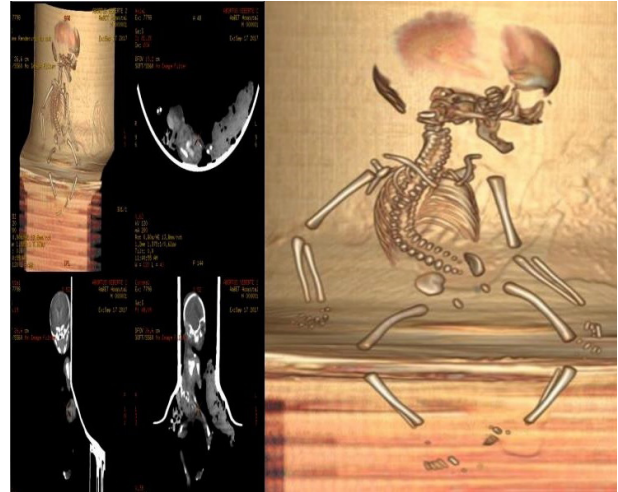
later the client was admitted for the therapeutic termination which was performed with induction of labor successfully, obtaining product of 20 weeks abortus weighing 200 gm and 8 inch long, with multiple and severe malformations.

The ultrasonography and MRI findings were confirmed. (Figure 9) with a good correlation between prenatal imaging findings and macroscopic findings for the Cantrell pentalogy. Taking consent from the parents; after radiography and CT was obtained (Figures 10,11) the abortus was sent for anatomico- pathological analysis.



Figure 9: Abortus of 20 weeks, lower thoracic and midline upper anterior abdominal wall defect with ectopia cordis and gastrichiasis (liver, stomach, spleen, and intestines out of the abdominal cavity, note cord

inserts on the left of the defect).



Figures 10,11: AP, LAT radiographs and 3D CT reconstruction of the abortus shows severe left scoliosis, clubbing deformity of the left foot bones. Note also the persistent extension at elbow with flexion deformity at wrist.

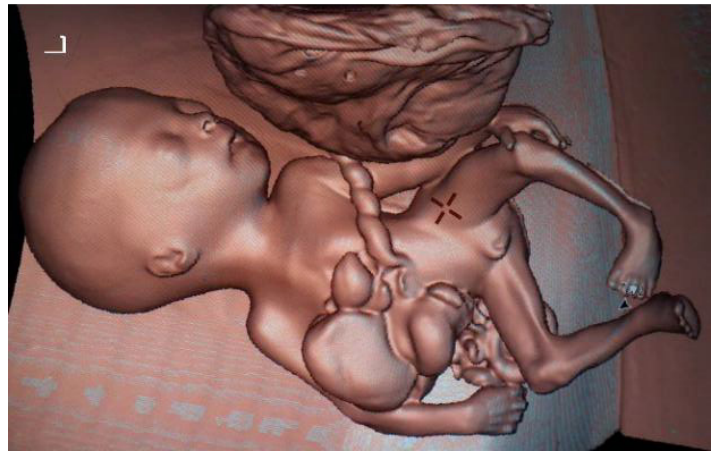


Figure 12: 3D CT with surface volume rendering shows a male abortus with ectopia cordis, gastrichiasis with a normal cord insertion on the right of the defect and clubbed left foot.



Discussion

The pentalogy of Cantrell is a rare and a severe congenital disorder characterized by the presence of ectopia cordis and an anterior abdominal wall defect was first described by Cantrell in 1958 [1]. Its incidence is very rare. Present in about 5.5/ 1,000,000 live births [2]. A combination of five congenital anomalies including supraumbilical midline epigastric abdominal defect, lower sternal defect, diaphragmatic pericardial defect, deficiency of the anterior segment of the diaphragm and congenital cardiac malformations make the diagnosis [1]. Estimated prevalence is almost three times higher in male compared to female [3]. The etiology and pathogenesis of pentalogy of Cantrell is unknown; although the developmental failure of a segment of the lateral mesoderm at 14-18 days of gestational age [1] and prevention of proper midline fusion of the chest wall due to rupture of the chorion and yolk sac is also suggested [5].

Kaplan LC genetic factors incriminated in the development of pentalogy of Cantrell includes mutation of BMP2 (bone morphogenetic protein 2) which is responsible for normal development of midline structures and ALDH1A2 which is important for conversion of vit A to retinoic acid which again plays a major role in organogenesis, pleuroperitoneal folding and in diaphragmatic embryogenesis [4,5]. Most cases of pentalogy of Cantrell are sporadic. Few cases associated with trisomy 18 and X linked inheritance have been reported [6].

Toyama classified pentalogy of Cantrell in to three different types in 1972.

Class 1. When all the five defects present.

Class 2. Four defects present including the ventral wall anomalies and intracardiac defects.

Class 3. An incomplete expression of this syndrome which always includes a sternal defect [7,8]. Sometimes malformations maybe so mild that making its discovery very difficult even after birth. Ectopia cordis often associated with this syndrome in 80% of cases [9].

Our case lies in class 1 with all five defects which identified on prenatal ultrasound, MRI , post mortem radiography and 3D reconstructed CT; finally clinical autopsy confirmed the imaging findings and also an associated atresia of the sigmoid colon.

The size and position of the anterior abdominal wall defect; its contents and association with other anomalies are features that are sought early in the diagnosis. Ultrasound is an invaluable tool for screening and diagnosis of pentalogy of Cantrell, Though 2D ultrasound is as equally efficient as 3D in an early stage of gestation [5]; using 3D ultrasound as adjuvant may help to enhance visualization of fetal anomalies in different orthogonal planes. [10]. Three dimension CT reconstruction is important in structural information, measurements and visualization of critical anatomic details. MRI offers an optimal assessment of this syndrome confirming and defining ultrasound findings and revealed supplementary diagnostic information with regard to the absent lower part of the sternum, the pericardium and the diaphragm [2]. Also it can be very helpful in cases where there is a severe oligohydramnios, where this prevents adequate visualization of the fetal parts using ultrasound [5].

There are also an associated severe left side scoliosis and clubbed foot which better depicted on CT in our case. Apart from these so many associated anomalies have been reported in literatures include: craniofacial and CNS anomalies such as cleft lip and palate, encephalocele, hydrocephalus, and craniorachisis (13) limb defects such as absence of tibia and radius, hypodactyly and even phocomelia [12,3].

Management of pentalogy of Cantrell is mainly depend on the extent of the defects and associated anomalies.

Most cases have a very poor prognosis which medical termination

of the pregnancy is advised. Surgical correction of the abdominal wall defect is often difficult in a complicated cases due to hypoplasia of the thoracic cage which makes incapable to enclose the ectopic heart [5]. But milder forms may be followed by a multidisciplinary team in order to determine the best time of delivery and corrective surgical treatments [3]. The results are obviously better in patients with minor defects [11].

Conclusion

Pentalogy of Cantrell is an extremely rare syndrome which should always be entertained when an ultrasound study reveals omphalocele and ectopia cordis. MRI, 3D ultrasound, and CT offers an optimal assessment of the syndrome by confirming and defining the 2D ultrasound findings. 2D ultrasound still remains the first mode of diagnosis in Pentalogy of Cantrell though these modalities give a supplementary diagnostic information, they don't change the prenatal counseling and the management plans. So here we emphasize on the importance of an early trimester antenatal ultrasound which allows early diagnosis of the syndrome, so that we able to appropriately counsel and manage the patient and minimize the psychological trauma to the patient.

Contributions

- 1: The author.
 - 2: Advisors.
 - 3: Did the clinical autopsy.
- Our gratitude: Eyuel Berihun.
- 4: For performing MRI scan.

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