Rarer expression of Pentalogy of Cantrell with Dandy Walker malformation and single umbilical artery
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Abstract:

Pentalogy of Cantrell (Thoraco abdominal ectopia cordis) is a rare congenital malformation syndrome consisting of a specific combination of ventral midline defects involving supraumbilical anterior abdominal wall, anterior diaphragm, diaphragmatic pericardium and sternum which can leads to various congenital anomalies like omphalocele and Ectopia cordis. Less than 90 cases have been reported in the literature. Here we report a case of incomplete Pentalogy of Cantrell with two rarer expressions of this disorder, Dandy walker malformation and 2 vessel cord which was not described in literature till now. Rarity of this entity is reason for presentation of this case report.

Key words: Ectopia cordis, Omphalocele, Pentalogy of Cantrell.

Introduction:

Pentalogy of Cantrell is a rare entity of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium and heart. The syndrome can be called as complete Pentalogy of Cantrell if it is associated with all five defects and incomplete if associated with three or four features in which presence of Omphalocele and ectopia cordis are hall mark of this syndrome.

Case Report:

Figure 1: ultrasound: Large Omphalocele.
A 24 years old second gravida (G2P1L1) with 5 months history of amenorrhea came for TIFFA (Targeted Imaging For Foetal Anomalies) to the Department of Radio diagnosis, Government General Hospital, Anantapuramu. She had caesarean section for previous pregnancy and had a 3 years old healthy child. Currently there are no histories suggestive of any medical problems like Diabetes, Hypertension and Thyroid disorders. There is no history related to intake of drugs or of consanguineous marriage.

Ultrasound scan (TIFFA) findings revealed single live intrauterine foetus of gestational age corresponding to 16-18 weeks as per parameters (Biparietal diameter, Head circumference – 18 weeks, Femur length – 16 weeks). Placenta was located at fundal region posteriorly with grade-0 maturity. Amniotic fluid was adequate for that gestational age. Multiple anomalies were detected which are as follows:

1. Ventral abdominal wall defect with large omphalocele (Fig 1). On thoracic examination, sternum and diaphragm were not seen and heart was located outside the thorax as a content of omphalocele (Fig 2A & B). Foetal cardiac anomalies were not evaluated due to herniation and unfavourable position of heart. Doppler showed four chambers of heart and foetal heart rate was good.
2. Moderate hydrocephalus with empty posterior fossa with non visualisation of Cerebellar hemispheres and vermis (Fig 3).
3. Two vessel Umbilical cord with single umbilical artery seen inserted at apex of omphalocele. (Fig 4).

Foetal spine, facial morphology and limbs were normal. With the above features of omphalocele, ectopia cordis, absent sternum and diaphragm, Pentalogy of Cantrell (Incomplete) was diagnosed with associated anomalies Dandy walker malformation and single umbilical artery. Overall the findings are suggestive of Pentalogy of Cantrell. The couple after counselling accepted for termination of pregnancy as Pentalogy of Cantrell with Ectopia cordis is a lethal congenital anomaly and is a definite indication for termination. Pregnancy was terminated with misoprostol and ended in complete abortion with anomalous foetus showing the above features. Specimen photographs (Fig 5) and foetograms were taken for demonstration of skeletal deformities. Foetograms showed absent sternum and empty thorax with herniation of heart into the Omphalocele (Fig 6). Foetal long bones appear normal.

Figure 2B: Colour Doppler: ectopic heart in Omphalocele

Figure 3: Ultrasound: moderate hydrocephalus, empty posterior fossa and non visualization of Cerebellar hemispheres and vermis
Discussion:

Pentalogy of Cantrell is a rare entity of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium and heart. It was first reported by Cantrell et al. in 1958 [1]. The incidence has been estimated to be 1 in 65,000-100,000 live births [2,3]. The aetiology of the Pentalogy is not well established. The proposed pathogenesis involves a defect in embryogenesis between 14 and 18 days after conception, when the splanchnic and somatic mesoderm is dividing. The sternum, abdominal wall, pericardium, and part of the diaphragm arise from somatic mesoderm, while the myocardium arises from splanchnic mesoderm. An event occurring prior to differentiation of the mesoderm into these two layers could produce defects in all of the involved structures, as seen in Pentalogy of Cantrell. The proposed

Figure 4: Ultrasound: Two vessel cord with single umbilical cord

Embryogenesis postulates a failure of the lateral mesodermal folds to migrate to the midline, causing the sternal and abdominal wall defects, and failure of the septum transversum to develop, causing defects in the anterior diaphragm and pericardium [4]. Congenital defects of the sternum may vary from simple notching of the manubrium to absence of the entire sternum. In our case complete absence of sternum observed.

Abdominal wall defects include Omphalocele, diastases recti, epigastric hernia, umbilical hernia, and combined defects. The most common abdominal wall defect is Omphalocele [2]. Deficiencies of the diaphragmatic pericardium and the anterior diaphragm are common defects. Cantrell et al..., stated that various congenital intracardiac anomalies are consistent elements of the Pentalogy, with ventricular septal defect is most common anomaly. Other congenital defects including head and facial deformities like meningocele, anencephaly, cleft lip, cleft palate, lung hypoplasia, adrenal aplasia, malrotation of the colon, hernia of the bowel into the pericardial cavity, undescended testicle, renal involvement and deformities of finger and foot are also described in literature.

Overall the prognosis appears dismal, but may be related to the extent of the defects. Toyama [5] demonstrated a survival rate of 20% in this disorder including its variants and incomplete syndromes. The complete Pentalogy has a poorer outcome, and the survival rate was only 8.5% in the report of Fernández et al [6]. If a diagnosis is made by ultrasound, then chromosomal analysis is recommended. Associations with trisomy 18, trisomy 13, and Turner syndrome have been reported. Careful imaging should be performed to rule out associated anomalies. Foetal echocardiography is indicated to evaluate the extent of any intra cardiac abnormalities. In view of the poor prognosis, termination of pregnancy may be considered if ultrasound diagnosis is made before viability.

Figure 5: Large hydrocephalus and Omphalocele

Figure 6: Foetogram: absent sternum and empty thorax
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