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CASE REPORT

ANTENATAL DIAGNOSIS OF PENTALOGY OF CANTRELL IN EAST AFRICA: A CASE REPORT

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Abstract

Introduction: The pentalogy of Cantrell is rare clustering of congenital defects, first described by Cantrell and colleagues in 1958. The exact pathogenesis for the pentalogy remains unknown and no specific genetic abnormalities have been correlated; however, a failure of embryogenesis has been suspected.

Case Presentation: We present a case of a 30 year old mother who had ante-natal ultrasonography which confirmed the presence of a single live fetus of 31.5 weeks gestation with severe oligohydramnios and the cardiac silhouette was protruding outside the chest cavity with no covering over the chest wall and no evidence of skin covering. There was no abdominal wall demonstrated and the abdominal contents appeared inhomogeneous with no normal position of the abdominal organs. In view of the ultrasound findings, the patient elected to have induction of labour and delivered by vaginal delivery. The fetus died within 30 minutes of delivery. Postnatal x-ray and autopsy were performed and this confirmed the antenatal ultrasound findings.

Discussion: Prenatal ultrasound diagnosis is possible as early as 10 to 12 weeks. 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 . Milder forms should be followed up by a multidisplinary team in order to determine the best time for delivery.

Conclusion: Pentalogy of Cantrell is an extremely rare congenital malformation who's exact aetiology is not completely understood. Antenatal detection of these anomalies is important since the prognosis is poorer in patients with complete forms of Pentalogy of Cantrell.

We demonstrate in this case report the extremely important role of early antenatal diagnosis of severe congenital abnormalities in these fetuses. By allowing us to diagnose these anomalies early, we are then able to appropriately manage the patient and minimize the psychological trauma to the patient.

INTRODUCTION

We wish to report the first case of Pentalogy of Cantrell in East Africa. Pentalogy of Cantrell is a rare syndrome first described by Cantrell in 1958 (Cantrell et al, 1958) . It is characterized by a spectrum 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 the lower sternum[1].Varying degrees of incomplete expression have been described and the defects range from subtle to severe.

CASE PRESENTATION

A 30 year old Gravida 2, Para 1, with one living child was referred to Moi Teaching and Referral hospital, Eldoret , because of a history of oligohydramnios at an outside ultrasound facility. She was dated at 32 weeks by last menstrual period. She and her husband were a non-consanguineous African couple with no family history of a hereditary disorder. There was no history of exposure to any teratogen. She neither took alcohol nor smoked cigarettes. Veneral Disease Research Laboratory test (VDRL) and Human Immunodeficiency virus test (HIV) were negative. Her medical and obstetric histories were unremarkable.

Antenatal ultrasound performed on an Esoate ultrasound machine confirmed the presence of a single live fetus of 31.5 weeks gestation with severe oligohydramnios. The cardiac silhouette was protruding outside the chest cavity with no covering over the chest wall and no evidence of skin covering. There was no abdominal wall demonstrated and the abdominal contents appeared inhomogeneous with no normal position of the abdominal organs. The spine was abnormal characterized with marked kyphoscoliosis at the thoraco-abdominal location. Despite the paucity of amniotic fluid volume a cleft palate was suggested. The antenatal diagnosis of Pentalogy of Cantrell was made based on the above sonographic features. In view of the ultrasound findings the patient elected to have induction of labour and delivered a 1500 gram fetus by vaginal delivery. The fetus died within 30 minutes of delivery. Postnatal x-ray was performed and this confirmed the antenatal x-ray findings of kyphoscoliosis of the spine. Gross macroscopic evidence of the fetus confirmed antenatal findings consistent with the diagnosis of Pentalogy of Cantrell. At autopsy the fetus also demonstrated total sternal agenesis, ectopia cordis which was seen sonographically as well as intracardiac defects were identified. Midline thoraco-abdominal wall defect with herniation of the intra-abdominal organs (omphalocele) was noted with absent anterior diaphragm and hypoplastic lungs were identified as were suspected from the antenatal ultrasound findings. Additionally, the fetus had a cleft face and lip which was also suspected antenatally. The left maxilla was not clearly identified. The left eye was missing. In addition, a shortened and malformed left upper limb was noted measuring 5 cm while the right upper limb was 14 cm long. There was a short and malformed left lower limb that was 10 cm long with one digit. The right lower limb was 6 cm long and the right foot had 4 digits. One hand was clenched with overlapping of the digits.The limb anomalies could not be assessed at the antenatal ultrasound because of the severe oligohydramnios. There were no external or internal reproductive organs identified.
Karyotyping done detected no aneuploidy for chromosomes 13, 18, 21 and sex chromosomes. The gender of the fetus was detected as female.
The above findings confirmed our antenatal diagnosis of complete Pentalogy of Cantrell which was described by Toyama et al (Toyama et al, 1972).

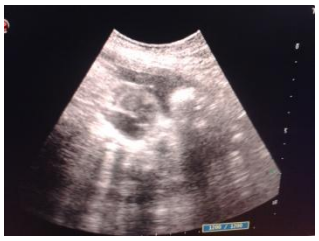

Fig 1: Fetal ultrasound: The visualised fetal heart

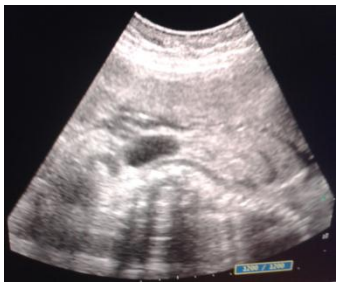
Figure 2: Fetal utrasound: Anterior thoraco-lumbar scoliosis with an anterior curvature

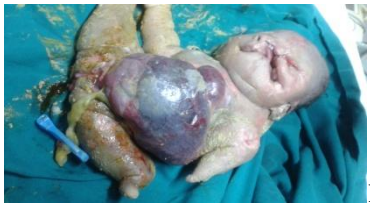
Figure 3: Absent anterior chest wall and abdominal wall.

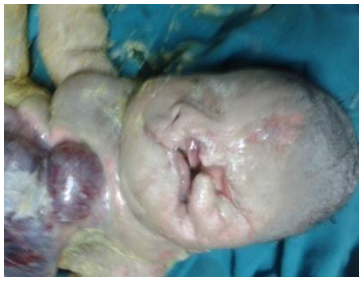
Figure 4: Cleft lip/palate extending into the left orbit & left nostril


Figure 5: Clenched hand with overlapping of the digits in the right upper limb

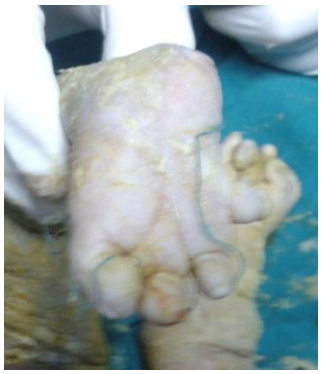
Figure 6: The right lower limb with four digits and a thick mid-foot plantar crease.

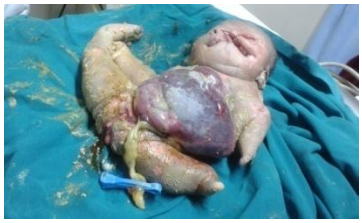
Figure 7: Left upper limb and left lower limb appear shortened on physical examination.

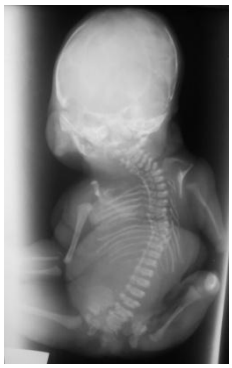
Figure 8: Post-natal Antero-posterior plain x-ray of the fetus: Depicts thoracolumbar kyphoscoliosis, Left upper limb only has a humerus and left lower limb only has a femur.

DISCUSSION

Pentalogy of Cantrell is a rare syndrome with an estimated incidence of 5 per million live births (Carmi et al, 1992) . Cantrell et al in 1958 first described this syndrome which is characterized by:
(1) midline supra thoraco-abdominal wall defect
(2) defect in the lower sternum
(3) deficiency of the diaphragmatic pericardium
(4) deficiency of the anterior diaphragm and various intracardiac abnormalities.
Sternal and abdominal wall defects cause herniation of the organs leading to (5) ectopia cordis and omphalocele (Cantrell et al, 1958).
Toyama suggested the following classification for the Pentalogy of Cantrell:
Class 1 = definite diagnosis with all five defects present.
Class 2 = probable diagnosis with four defects present including intracardiac and ventral wall anomalies.
Class 3 = incomplete expression with various combinations of defects including a sternal abnormality[2].

The etiology of Pentalogy of Cantrell is unknown; however, the longstanding suggested mechanism for the development of Pentalogy of Cantrell has been the developmental failure of a segment of the lateral mesoderm at gestational age 14 to 18 days. As a consequence, the paired mesodermal folds of the upper abdomen do not migrate ventro-medially and the transverse septum of the diaphragm does not develop [1].Most cases are sporadic and no recurrences have been reported. There is a male dominance with the male to female ratio of 2.7 to 1 (Cantrell et al, 1958). Based on embryological development, this syndrome may be classified into two groups. The first group which is composed of three of the defects, that is, diaphragmatic defect, pericardial defect and intracardiac lesions which arise as the result of developmental failure of a segment of the mesoderm. The second group includes the sternal and abdominal wall defect and appears to arise due to failure of migration of the paired primordial structures (Cantrell et al, 1958). Varying degrees of incomplete expression have been described according to the postulated embryological development of these defects (Cantrell et al, 1958). (Sanchis et al, 1992), (Chen et al, 1997).

Intracardiac anomalies described in the Pentalogy of Cantrell include septal defects, Ebstein's anomaly, Tetralogy of Fallot's or single atrium. In 75% of the cases, pericardium may be absent (Cantrell et al, 1958), (Morales et

al,2000), (Vasquez-Jimenez et al, 1998). In our case the 4 chamber view was markedly abnormal but an exact congenital heart diagnosis could not be attained.

Other associated anomalies have been reported including craniofacial and central nervous system anomalies such as cleft lip and palate as demonstrated in the fetus that we have described, encephalocele, hydrocephalus and cranio-rachischis (Morales et al, 2000), (Correa-Rivas et al, 2004), (Polat et al, 2005). Limb defects such as club foot, absence of tibia or radius, hypodactyly or even phocomelia (Pivnick et al, 1998), (Uygur et al, 2004), (Sunil et al, 2014) are also common as was seen on the post-mortem findings in the case report that we have described. Abdominal organ defects such as gallbladder agenesis and polysplenia, may also be present (Bittmann et al, 2004).

Antenatal diagnosis can be made as early as 10 to 12 weeks. The earlier this diagnosis is made in pregnancy the easier it is to evaluate all the phenotypic abnormalities due to the fact that the amniotic fluid is not as severely diminished in the early part of the gestation. The size and position of the anterior abdominal wall defect, its contents and its association with other anomalies are features that are sought early in the diagnosis. Using 2D ultrasound in the first and second trimester as well as a 3D ultrasound as an adjuvant, may help enhance visualization of fetal anomalies in different orthogonal planes (Rodgers et al, 2010). Fetal chromosomal analysis is recommended if a diagnosis is made by ultrasound since there have been reported associations with Trisomies and Turner syndrome (Morales et al, 2000).

Termination of pregnancy may be offered in severe cases and especially when amniocentesis reveals an abnormal karyotype. The earlier this is performed in a patient the less emotionally traumatic it is to the mother. Preparing her for this type of delivery antenatally will facilitate and minimize the psychological distress to the patient.

Milder forms may be followed by a multi disciplinary team in order to determine the best time of delivery (Suresh et al, 2013).

CONCLUSION

Pentalogy of Cantrell is an extremely rare congenital malformation whose exact cause is not completely understood. The complex has variable clinical expression with complete and incomplete expressions reported. In our case, it was associated with other anomalies. Antenatal detection of these anomalies is important since the prognosis is poorer in patients with complete forms of Pentalogy of Cantrell, ectopia cordis and patients with associated anomalies. We demonstrate in this case report the extremely important role of early antenatal diagnosis of severe congenital abnormalities in these fetuses. By allowing us to diagnose these anomalies early, we are then able to appropriately manage the patient and minimize the psychological trauma to the patient. In cases which are not incompatible with life a multidisciplinary approach is essential to optimize successful management of these patients (Suresh et al, 2013).

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