Case Report

Case Report: Pentalogy of Cantrell with Cranioschisis and Facial Abnormality; An Extremely Rare Association

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ABSTRACT

Pentalogy of Cantrell is a rare multiple congenital anomalies syndrome that is characterized by 5 major defects. The defects include midline supraumbilical abdominal wall defect (omphalocele), lower sternum defect, anterior diaphragm defect, diaphragmatic pericardium defect and heart defect (ectopic cordis and intracardial defect). In extreme cases, the condition is not compatible with life. The prognosis is even worse when it is associated with other complex anomalies. The exact cause is not completely understood. It occurs sporadically in the majority of cases with variable clinical expressions, though it has been linked to some chromosomal anomalies such as Trisomy 18 and some X-linked disorders. Complete and Incomplete expressions have been reported. We hereby report a case of incomplete manifestations of Pentalogy of Cantrell with rare associations (Cranioschisis, cleft lip and palate). The challenges encountered in making diagnosis (both prenatally and postnaschisis, tally) as well as treatment are hereby discussed.

Keywords: Association, Cantrell, CranioFacial anomaly, Pentalogy.

INTRODUCTION

Pentalogy of Cantrell(POC) is a rare congenital malformation. Cantrell in 1958 described the full spectrum of the Pentalogy to consist of 5 major defects. These include midline supraumbilical abdominal wall defect, defect of the lower sternum, deficiency of anterior diaphragm, diaphragmatic pericardium defect and various congenital and intra-cardiac abnormalities. [1,2]The prognosis is worse when all the components are present. However, not all patients present with all the components.[3] Ectopic cordis is a typical feature of Pentalogy of Cantrell defined as partial or complete displacement of fetal heart outside the thoracic cavity. Its prevalence is 7.1 per million of live birth.[4]Four types have been identified, namely: cervical ectopic cordis (3% of all cases), thoraco-abdominal ectopic cordis (7% of all cases), abdominal ectopic cordis (30% of all cases), thoracic ectopic cordis (60% of all cases).[5] Other structural anomalies that may occur in association with Pentalogy of Cantrell include craniofacial defects such as cleft palate and supranumeral nares, nervous system defect such as hydrocephalus, encephalocele, anencephaly and neural tube defect. Besides, skeletal defect (club foot) and abdominal anomaly like malrotation of the colon may occur.[6,7]

Toyama grouped Pentalogy of Cantrell into 3 classes; Class I: Occurrence of all 5 defects, Class II: Occurrence of 4 defects with intra-cardiac and

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ventral abdominal wall defect present and Class III: Incomplete expression showing various combinations of defect with definite presence of sternal anomalies.[8]

It has a worldwide distribution with a reported incident of 5.5 per million live birth. [1] Some authors have also reported the prevalence of the syndrome to vary from 1:65,000 to 1:200,000.[9]

The exact cause is unknown but however Cantrell *et al* when describing the syndrome attributed the anomaly to developmental failure between 14 to 18 days of embryonic life, involving an inappropriate differentiation of the lateral mesoderm.[2] It has also been attributed to some chromosomal anomaly like trisomy 18 and Goltz-Gorlin Syndrome [10,11] as well as some X-linked disorders. [12]

Like in other parts of the world, the prevalence of Pentalogy of Cantrell is rare in Nigeria. To the best of our knowledge, this case is the 5th series of cases of Pentalogy of Cantrell reported in Nigeria. [13-16] Most of the cases were incomplete Pentads and none was associated with craniofacial anomalies.

We report an incomplete pentad with cranioschisis and facial anomalies. The challenges of prenatal and postnatal investigations as well as treatment in our setting as our report is based on gross features.

CASE REPORT

A female neonate was admitted from birth with multiple congenital anomalies- Omphalocoele, complete cervical ectopic cordis, lower sternal defect, anencephaly, encephalocele, cleft lip and palate. She was delivered via emergency caesarean section to a 28-year -old un-booked now Para 1 + 2 (2 previous spontaneous miscarriages) woman at term. Indication was intrapartum hemorrhage in labour and multiple gestation.

She was first seen in our facility in labour with an ultrasound done earlier in the day at a peripheral centre which revealed multiple gestation. Her pregnancy was booked at a Primary Health Centre in town at gestational age of 5 months. Subsequent antenatal visits were uneventful; had her routine haematinics with two obstetrics Ultrasound scan done in the course of pregnancy, which were both adjudged to be normal. She neither smokes nor drinks alcohol. No history to suggest the use of teratogenic agents or un-prescribed medications. Not a known hypertensive or diabetic and not on any routine drug for chronic medical condition. However, has had 2 previous spontaneous miscarriages.

She is a Seamstress married in a non-consanguineous union to a 30 year old artisan in a monogamous setting. Examination revealed a female neonate with features of term, weighing 3.1kg, had no cranium but with brain materials floating on the frontal and occipital regions, absence of eyes but with a pair of nares, no nasal bridge, there was presence of both cleft lip and palate. There was a hypoplastic heart located in the right aspect of lower neck and anterior thorax which was actively pulsating. The heart was attached proximally to the forebrain and distally to the liver (Figure 1).

There was a lower sternal defect. The liver and part of the guts were all enveloped in a thin membranous sac (without skin) attached to the umbilical stump visible at the defect on the supra-umbilical anterior abdominal wall. The Omphalocoele measured 7cm x 6cm. (Figure 2). Bowel sounds were present and baby had passed meconium. Baby was grunting, tachypneic and tachycardic, cyanosed peripherally. No gross spinal or limb deformities. Chest x-ray and abdominal ultrasound was not done because father declined investigations and treatments due to financial and emotional constraints. Echocardiography could also not be done for similar reasons and the fact that our machine has no probe for neonatal studies.

A diagnosis of Pentalogy of Cantrell of at least class III with cranioschisis, cleft lip and palate was made based on gross features due to our limitation in carrying out imaging studies to decipher intracardiac and diaphragmatic abnormalities. Parents were duly counseled on baby condition and possible outcome. Baby died about 12hrs into admission.

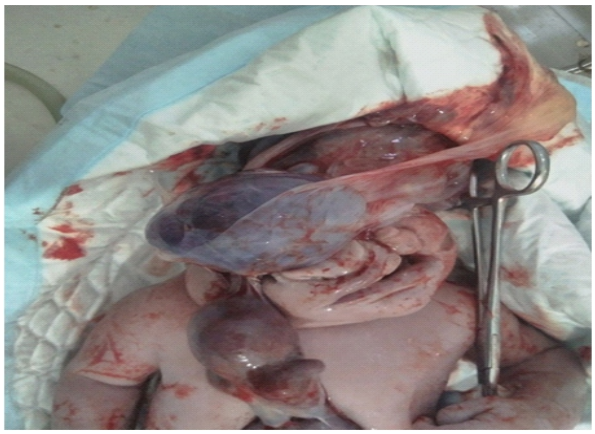

Fig. 1

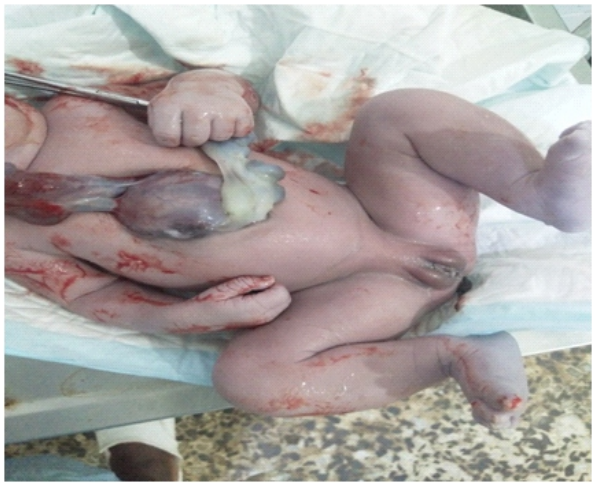

Fig. 2

DISCUSSION

The Pentalogy of Cantrell is an extremely rare phenomenon and even rarer with craniofacial abnormalities with a prevalence of 1:65,000 live births.[17- 19] It is commoner in boys and less than 60 cases have been documented globally as at 2007.[17] To the best of our knowledge, this case is the 5th series of cases of Pentalogy of Cantrell reported in Nigeria[13-16] and the first in our Centre. However, our patient was a female. Most of the cases were incomplete Pentads and none was associated with craniofacial anomalies. The case we reported was a case of incomplete pentad but it is unique in that it is associated with craniofacial anomalies.

The pathogenesis of POC is not fully understood. Cantrell*et al* [2] suggested an embryologic developmental failure of the segment of the lateral mesoderm around the gestational age of 14 to 18 days. Consequently, the transverse septum of the diaphragm does not develop and paired mesoderm folds of the upper abdomen do not migrate ventromedially. Organs may eviscerate through the sternal and abdominal wall defect. Though different theories have been proposed to explain the etiology of this condition, such as the intrauterine drug exposure. In our case however, mother denied the use of any drug during pregnancy. The disorder varies in severity from individual to individual. Majority die shortly after birth in severe cases especially when associated with cranioschisis and intracardiac defect. However, a 32-year-old man has been reported to be living with the condition.[20] Our patient had extremely rare and severe association of cranioschisis and facial anomalies with the Toyama class 3 disorder and died about 12 hours after delivery.

POC can be diagnosed prenatally within the first trimester.[21] This has not been the pattern in cases reported in Nigeria due to limited number of centres with facilities for fetal anomaly scan. Even though in our case, mother had two prenatal scan, none of them was able to diagnose the condition and sadly one of the two ultrasound scan showed twin gestation. This underscores the limitation centers that are able to do fetal anomaly scan, and further emphasizes that the diagnosis depends on the sonographers' expertise of fetal ultrasound. POC has array of associated intra-cardiac defect (VSD100%, ASD 53%, and Ventricular Diverticulum 23%.)[2] In addition to the aforementioned intra-cardiac defect that have been reported previously in patient with ectopic cordis described in the literature, other anomalies have been reported in patient with ectopic cordis such as trisomy 18, cleft lip and palate, Neglected Tropical Diseases (NTD), genitourinary anomaly, pulmonary hypoplasia, liver enviseration.[22] Our patient in addition to other features earlier mentioned had cleft lip and palate and ectopic liver which concurs with studies reported in the literature.

After birth, echocardiography is essential for diagnosis of other cardiac anomaly. Other features and associated anomalies can be diagnosed by conventional radiography or sonography. Nevertheless, small defects of the diaphragm and pericardium can be extremely difficult to diagnose accurately.[23, 24] MRI and CT Scan may also be useful. However, in our setting further evaluation of a neonate with such anomaly after adequate counselling of the parents was difficult because they declined consent for further evaluation and care due to socio-cultural belief, financial and emotional constraints.

CONCLUSION

Pentalogy of Cantrell is generally less lethal in incomplete expression. However, the severity of intra-cardiac lesion and associated anomalies tend to worsen the prognosis. Little wonder did the reported case died. Even though it was a case of Incomplete Pentad, there was associated cranio-facial anomaly and suspicion of intra-cardiac defect. Despite the increasing awareness about fetal anomaly scan in developed countries, prenatal diagnosis is still a major challenge in our environment. Poverty, cultural and ethnic beliefs also make postnatal evaluation difficult in our setting. The importance of fetal anomaly scan during antenatal period should be stressed among the stakeholders and such information should be incorporated into the health education package during routine antenatal visits.

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