Case Report

Pentalogy of Cantrell in Ile-Ife: A Case Report and Review of the Literature

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ABSTRACT

Pentalogy of Cantrell is a rare congenital malformation. The aetiology is not well understood, but genetic and sporadic forms have been reported. It is a combination of five congenital defects: the heart, pericardium, diaphragm, sternum and anterior abdominal wall. Only a few cases of pentalogy of Cantrell have been reported in Nigeria. Prenatal diagnosis is possible at the beginning of the second trimester using ultrasonography. Success in the management of this condition requires expertise and a multidisciplinary approach. We report the case of a 7-day-old boy, who was referred to our hospital in Ile-Ife with a chest wall defect at birth and ectopia cordis, diaphragmatic hernia seen on ultrasound and multiple heart defects seen on echocardiography. The diagnosis of pentalogy of Cantrell was made. The severity of the cardiac defects in this case and the late referral contributed to the poor outcome of the reported case. Other difficulties in the management of this case peculiar to our environment are highlighted.

Key words: Cantrell, ectopia cordis, Ile-Ife, pentalogy

How to cite this article: Ugowe O, Afeniforo O, Adeyefa B, Okeniyi JA, Adejuyigbe EA. Pentalogy of cantrell in Ile-Ife: A case report and review of the literature. Niger J Health Sci 2019;19:81-4.

INTRODUCTION

Pentalogy of Cantrell was described by Cantrell *et al*.[1] in 1958 after reporting five cases of children with ectopia cordis associated with diaphragmatic hernias, partial pericardium, defects in the ventricular septum and a part of the sternum. It is a rare condition with an estimated incidence between 1 in 65,000–200,000 live births. The aetiology is not exactly known, however, Cantrell suggested that a failure in the development of the lateral mesoderm resulting in poor migration and differentiation, occurring at 14–18 days of embryonal life, is responsible for the pentalogy.[1-3] Abdominal or thoracic organs may herniate from the defect. Three classes of this malformation exist with variety in presentation.[4] Ectopia cordis may be partial or complete. Ectopic locations of the heart may include the thoracic, abdominal or even the cervical region. The great vessels may be exposed.[4]

A case was reported in Kano by Mukhtar-Yola *et al*.[5] in 2012, a full-term neonate with ectopia cordis, diaphragmatic and sternal defects with an absent pericardium. Three cases were previously reported by Sowande *et al*.[6] in Ile-Ife. Ages at presentation were 18 h, 36 h and 6 weeks, respectively. Aliyu and Mohammad[7] also reported a case in a 9-month-old infant in Kano. Okafor *et al*.[8] and Pius *et al*.[9] reported variants of pentalogy of Cantrell in Enugu and Maiduguri, respectively. Prenatal diagnosis is possible as early as 13–14 weeks of gestation.[10] Management is often challenging because of the high level of expertise needed and the complexities of the defects. Factors that result in poor outcome for children with pentalogy of Cantrell include late presentation and the presence of severe cardiac malformations. This case is being reported because the true prevalence of this anomaly is not known in our environment and some local studies have only reported variants of this condition.[8,9] We hereby report a case of pentalogy of Cantrell in a 7-day-old male who weighed 3.29 kg. Our patient had a Class 1 defect because all components of pentalogy of Cantrell were represented. We also highlight the challenges we faced in the management of the baby.

Submitted: 08-Sep-2020 **Revised:** 30-Sep-2020
Accepted: 26-Dec-2020 **Published:** 30-May-2022

Access this article online

Quick Response Code:

Website: www.chs-journal.com

DOI: 10.4103/njhs.njhs_45_20

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CASE REPORT

The patient (Baby A) was a preterm male neonate, born at 36-week gestation who weighed 3.29 kg and was admitted at 7 days of life in the neonatal ward of Obafemi Awolowo University Teaching Hospitals Complex, Ile-Ife, Osun State, Nigeria. The baby was referred from Olabisi Onabanjo University Teaching Hospital (OOUTH), Sagamu, Ogun State, after 6 days of admission there for cardiothoracic review and further management. He presented with an anterior chest and abdominal wall defect noticed from birth with a protruding mass which had a thin skin covering and visible pulsations. He was also noticed to have bluish discoloration of the lips and mucosa at presentation. There were no vomiting and no abdominal distension, and he had moved bowel regularly.

The baby was delivered via spontaneous vaginal delivery in a private hospital in Ogun State. The mother booked pregnancy at about 20 weeks in the same private hospital where she also had antenatal care. Prenatal ultrasound done did not reveal any congenital anomaly. There was no fever with rash during the first trimester, no use of unprescribed medications or herbal concoction during the pregnancy and no exposure to ionising radiation. She neither smokes cigarette nor takes alcohol. At 6 months into the pregnancy, the mother had preterm contractions and was admitted and managed for 2 weeks as a case of threatened abortion. However, she had preterm contractions and delivered at 36 weeks + 1 day. He is the third child out of three children in a monogamous family setting. The other siblings are females and they are alive and well.

The mother is a 33-year-old, para 4 + 1 woman whose second parous experience resulted in still birth. The mother did not volunteer information about the circumstances surrounding the still birth. There was no history to suggest consanguinity. The father is a 37-year-old fish farmer, educated up to senior secondary school level, while the mother is a primary school teacher, also educated up to senior secondary school.

On general physical examination here in Ife, he had poor activity, was not pale and had a normal temperature of 36.8°C. He was centrally cyanosed with respiratory distress as evidenced by flaring of alar nasi and intercostal recessions. The weight at admission was 3.29 kg, while the length and occipitofrontal circumference were 48 cm and 33 cm, respectively, all within normal range. There were no dysmorphic facies. On systemic examination, he had an anterior thoracoabdominal defect which measured 10 cm by 8 cm, extending from just above the xiphisternum to the lower epigastrium, with the exposed heart covered by dry, hyperpigmented and ulcerated skin [Figure 1]. The heart was covered by the ulcerated skin and located in the epigastric region with visible heart pulsations. The great vessels were not exposed, and the umbilicus was intact. On cardiovascular examination, the heart rate was 142 beats/min; there was a systolic murmur heard over the heart. The respiratory rate was 56 cycles/min. There were crepitations heard all over the lung fields, especially posteriorly. Oxygen saturation at our centre was 55%, compared to a saturation of 85% in OOUTH, Sagamu, where the baby previously spent 6 days on admission was 85%. On genital examination, it was found that the scrotal sac was empty, but testicles were palpable in the inguinal region. On musculoskeletal examination, no lumbosacral swelling was observed.

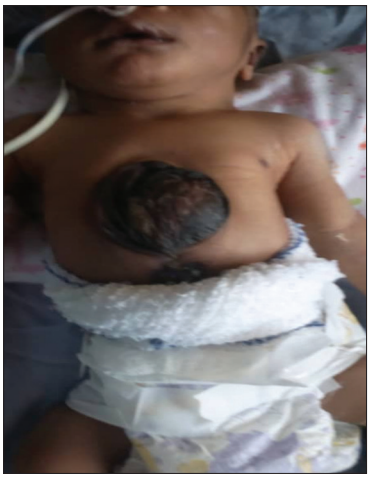

Figure 1: An epigastric mass covered by a hyperpigmented sac

The baby had a packed cell volume of 49%, while the random blood sugar, electrolytes and full blood count were within normal limits. Chest radiogram showed bowel loops in the chest [Figure 2]. Two-dimensional echocardiography revealed multiple complex congenital defects which included a pericardial defect, large ostium secundum type of atrial septal defect (ASD), multiple muscular ventricular septal defect (VSD), tricuspid atresia, dilated coronary sinus and supracardiac type partial anomalous pulmonary venous drainage [Figure 3]. Ultrasound demonstrated a thoracic defect with a diaphragmatic hernia and bowel loops around the heart. The diagnostic features of pentalogy of Cantrell seen in this baby were sternal and abdominal wall defects, pericardial and diaphragmatic defects seen on echocardiography and ultrasound respectively and an abnormally positioned heart (ectopia cordis) with multiple cardiac defects.

He was commenced on intravenous fluids 8.3% dextrose saline, intravenous ceftriaxone and gentamicin. He was placed on intranasal oxygen. Oxygen saturation ranged from 47% to 55%. The exposed thoracoabdominal sac was cleaned daily with normal saline and covered with Sofra-Tulle™. The condition and prognosis of the patient were explained to the mother. The paediatric surgeons, paediatric cardiologists and cardiothoracic surgeons were involved. The paediatric surgeons aimed to have a staged surgery where the initial stage was to close the diaphragmatic defect to allow for the cardiothoracic surgeons to operate at the nearest available date. This is to allow for preparation for cardiac surgery. The oxygen saturation of the patient, however, did not exceed 55% at any time during the admission. This made the baby a high risk for surgery and pre-operative anaesthesia, as highlighted by the paediatric surgeons.

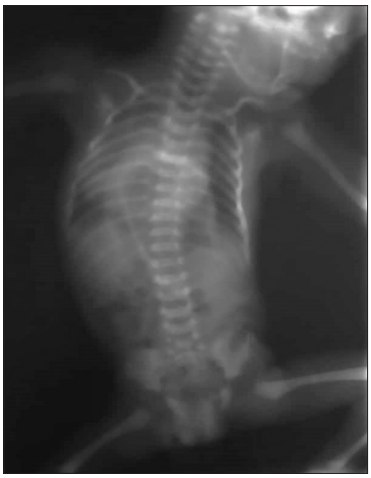

Figure 2: A plain babygram

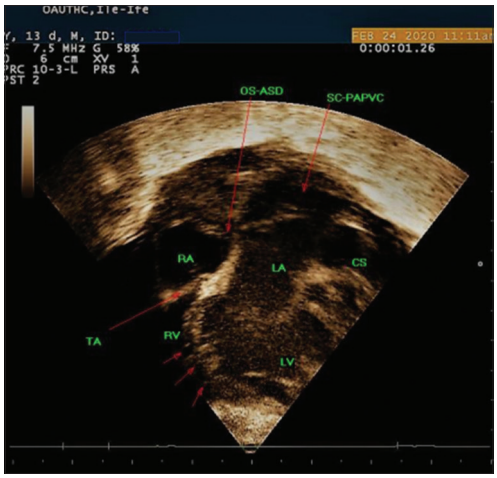

Figure 3: Echocardiogram. CS: Dilated coronary sinus, LA: Left atrium, LV: Left ventricle, RA: Right atrium, RV: Right ventricle, OS-ASD: Ostium secundum-type atrial septal defect, SC-PAPVC: Supracardiac chamber-type partial anomalous pulmonary venous circulation, TA: Tricuspid atresia and the arrows depict multiple muscular ventricular septal defects

On the 5th day of admission, he had apnoea which lasted for about 2 min after which he resumed spontaneous respiration after Ambu bagging. He had about three episodes of apnoea over the next 24 h. On the 6th day of admission, he had another episode of apnoea and all resuscitative measures were not successful and he was declared clinically dead.

DISCUSSION

Pentalogy of Cantrell is a combination of five congenital defects which are defects of the heart, pericardium, diaphragm, sternum and anterior abdominal wall, including a heart that is totally or partially located outside the thorax (ectopia cordis). An omphalocele found in 74.5% of patients is the most common abdominal wall malformation. This is followed by defects of the lower sternum (59.4%), diaphragm (56.8%) and pericardium (41.8%).[1,5] Pentalogy of Cantrell is due to a developmental defect of the mesodermal layer occurring at 14–18 days post-conception.[1,10] This leads to migration of the visceral mesoderm over the abdomen to the chest leading to the abnormal formation of the sternum and abdominal wall.[10] Toyama[11] described three classes of Cantrell's pentalogy in 1972. Class 1 has all five defects present. Class 2 has only four defects present, including intracardiac and ventral wall abnormalities as essential components. Class 3 is an incomplete expression with a combination of defects, with sternal defect being an essential component. Our patient belonged to the Class 1 group in which a definitive diagnosis can be made.[4,11]

Cantrell described the most common heart lesions to be a VSD occurring in all cases, ASD occurring in about 53% of cases and pulmonary stenosis or atresia occurring in 33% of cases[1,12] More recent evidence has shown that VSDs are not universally present but occur in 72% of cases.[13] Our patient had an ectopia cordis with echocardiography diagnosed ventricular and ASDs, which conforms to the commonly encountered cardiac defects in pentalogy of Cantrell. Unlike in our case where echocardiography was done, the cases reported by Mukhtar-Yola *et al*.[5] in Kano and Sowande *et al*.[6] in Ife did not have echocardiography done due to unavailability of paediatric echocardiography services. However, the complexity of the cardiac defects in our index case needed further evaluation using cardiac catheterisation, a procedure which the parents could not afford. Other associated anomalies that were absent in the index case but featured in other reported cases of pentalogy of Cantrell include cleft lip/palate, hydrocephalus, spinal defects and limb defects.[5,12,13]

The index case had a prenatal ultrasound done in a private hospital that showed no abnormality. The parents were surprised to see such myriad of defects. This is sadly a recurring phenomenon in our environment where there is a lack of expertise in making a prenatal diagnosis using ultrasound. In the Mukhtar-Yola *et al.* report,[5] the mother did not have an ultrasound done throughout pregnancy. All three cases reported by Sowande *et al*.[6] in Ife also did not have any form of prenatal diagnosis. Making a correct prenatal diagnosis is largely dependent on the proficiency of the sonographer as well as the quality of the machine used. Training of personnel to improve competence in detecting congenital anomalies in early pregnancy will go a long way in reducing the incidence of missed diagnosis. Pentalogy of Cantrell can be diagnosed as early as 14 weeks, where the facilities and expertise exist. Features that can be seen on ultrasound include midline abdominal defect, ectopia cordis and cardiac malformations.[14] Early diagnosis will ensure effective planning by the specialists involved (Obstetricians, paediatricians and paediatric and cardiothoracic surgeons) and preparation for delivery and subsequent surgery.

Our patient also maintained a very low oxygen saturation due to the existing cardiac defects. An earlier referral on the day of birth, when oxygen saturation was still above 80% as reported in the referral note, may have allowed for an earlier

repair of the diaphragmatic defect. The paediatric surgeons expressed concern that the baby may not survive pre-operative anaesthesia, a viewpoint that was also echoed by the intensivists. Alexis[15] reported a case of a 2080-g baby born in South Africa with pentalogy of Cantrell that had midline defects closed at surgery. The oxygen saturation was normal before surgery probably because the heart defects diagnosed were an ASD and a small patent ductus arteriosus, unlike our index case who had an ASD, VSD and a right outflow tract obstruction.

Management of pentalogy of Cantrell is mainly surgical with a relatively high rate of mortality, as high as 37%, even where adequate facilities exist.[13,16] Balderrábano-Saucedo *et al*.[16] reviewed outcome of 22 cases of pentalogy of Cantrell. Mortality rate recorded was 73%, with two-thirds occurring in babies with ectopia cordis. Surgery usually consists of corrective or palliative cardiovascular surgery, correction of diaphragmatic hernia and correction of any associated anomaly. The type of surgery to be done depends on the severity of the heart defects, the presence of ectopia cordis and the size of the abdominal wall defect.[12,17] Surgery can be single staged or multistaged. The single-staged procedure is mainly for patients with less severe cardiac defects, while in a multistaged procedure, correction of intracardiac abnormalities is given preference before surgery on defects of the thoracoabdominal wall is carried out.[12] An initial diaphragmatic repair was proposed for our patient because cardiac surgeries are usually scheduled and not routine, due to the extensive preparedness and logistic issues associated with carrying out cardiac surgeries in our environment. If the cardiac malformations of our index case had not been severe, the baby may have lived long enough to have diaphragmatic repair and scheduled cardiac surgery. However, the experience in a few centres in Nigeria where Cantrell's pentalogy has been reported is that most parents discharge their babies against medical advice due to financial constraints, even before surgery is done.[5,6,8]

CONCLUSION

We are still limited in our ability to recognise congenital malformations prenatally because of lack of widespread facilities and expertise. When pentalogy of Cantrell is suspected, a multidisciplinary team involving an obstetrician, neonatologist, paediatric surgeon, paediatric cardiologist and cardiothoracic surgeon should be mobilised on time to prepare for the arrival of the baby. These measures may, however, be limited in our setting because of late presentation and poverty. Prenatal diagnosis of congenital malformations is still problematic in our environment, and prenatal diagnosis will enable adequate multidisciplinary preparation for such patients as well as improve survival.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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