THE COMPLEX ANOMALY OF THORACO-ABDOMINAL ECTOPIA CORDIS IN IBADAN: MAKING A CASE FOR IMPROVED NHIA COVERAGE: A CASE REPORT

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ABSTRACT

Background: **Pentalogy of Cantrell (POC) is a collection of five congenital malformations involving the heart, pericardium, diaphragm, sternum, and anterior abdominal wall. This is a spectrum, and could be incomplete or complete; naked or covered cordis or can be classified based on the location of the heart, and in this index patient – thoraco-abdominal ectopia cordis. We aim to highlight this finding of covered incomplete pentalogy of Cantrell and the peculiarities of management in our sub-region that relies solely on out of pocket payment.**

Case presentation: **A 30-hour old female admitted with history and clinical examination findings in keeping with a covered pentalogy of Cantrell (Thoraco-abdominal ectopia cordis) in a term neonate. Attached is a video link at presentation https://youtu.be/0vQuzq14G9Q**

Conclusion: **Pentalogy of Cantrell is a complex anomaly. The constraints we encountered in the initial management of this neonate is not uncommon in surgical practice within Nigeria, and other resource constrained countries especially amongst carers of paediatric patients. Other factors at play in our sub-region are ignorance, high rate of illiteracy, superstitious beliefs, and varied cultural practices, and this partly hindered further evaluation of this vulnerable neonate in our sub-region.**

Keywords: Complex anomaly, Ibadan, Pentalogy of Cantrell, Thoraco-abdominal ectopia cordis

INTRODUCTION

The pentalogy of Cantrell (POC) was first described in the mid-20th century.[1] It is also known as Cantrell-Haller-Ravitch syndrome[2] with a collection of congenital malformations involving the heart, pericardium, diaphragm, sternum, and anterior abdominal wall.

The cardinal features of the syndrome were anterior abdominal defect (omphalocele) in association with ectopia cordis (EC). Toyama (1972) subsequently described the full or complete spectrum of the Cantrell's syndrome to consist of ectopia cordis, a defect in the diaphragmatic pericardium, a deficiency of the anterior diaphragm, lower sternal defect, a midline supraumbilical abdominal wall defects, various structural congenital intracardial anomalies.[3] The incomplete (partial) refers to the presence of less than five of the defects.

We present a case of incomplete pentalogy of Cantrell with covered heart however, patient's care givers discharged against medical advice to seek for traditional medical care due to cost of investigations and care. This further exemplifies of the problems of managing paediatric patients with congenital anomalies in our sub region where majority pay out of pocket, and this report is a clarion call for us to do more in this regard.

Case Presentation

A 30-hour old female was brought to the neonatal emergency room by parents after delivery in a peripheral centre with an anterior abdominal wall defect noticed at birth. No features of intestinal obstruction.

She was delivered to a 28-year-old P_2^{+0} woman via spontaneous vaginal delivery and delivery was supervised by an unskilled staff. The child however, cried well at birth with a birth weight of 2.8 kilograms. No history suggestive of exposure to teratogens during pregnancy, mother though not regular with antenatal clinic visits however, claimed to have taken folic acid amongst other haematinics during pregnancy. No family history of similar condition. Other sibling is alive and well.

Examination showed a term female with macroglossia, pink in room air, and not cyanosed. (Figure: 1). The anterior fontanelle was patent and normotensive with appropriate head circumference.

Chest examination showed a respiratory rate of 60 / minute, a lower sternal defect with visible cardiac impulse within a thin membrane covering the epigastric defect. Attached is a video link https://youtu.be/0vQuzq14G9Q

There was no respiratory distress, she had normal breath sounds bilaterally. Heart rate was 164 / minute, heart sounds I and II were heard, and no murmur.

Figure 1: A term female neonate with macroglossia, and covered POC

The abdomen moved with respiration. There is an epigastric defect with its widest diameter measuring seven centimeters, and the umbilical cord stump normally sited. (Figure: 2)

Figure 2: Epigastric omphalocele with umbilical cord stump on the midline.

Perineal examination revealed normal female external genitalia with patent and normal anus. No limb or vertebral defects seen.

A diagnosis of covered Pentalogy of Cantrell (Thoraco-abdominal ectopia cordis) in a term female neonate was entertained.

Dextrose containing intravenous fluid was commenced with normal glycaemic control maintained.

Packed cell volume is 50%. Echocardiography was deferred due to financial constraints at presentation, and did not afford us the opportunity to rule out intracardiac defect.

We however, commenced daily dressing of the membranous covering with 1% silver sulphadiazine, Sofra-tulle, and layer of sterile gauze.

Parents discharged the neonate against medical advice about 4 hours into admission, due to financial constraints, cultural belief about the cause of the anomaly as they opted for traditional medical care, and was lost to follow-up.

DISCUSSION

Pentalogy of Cantrell (POC) was first described by Cantrell, *et al.* in 1958, based on the presence of a midline supra-umbilical abdominal wall defect, a defect of the lower sternum, deficiency of anterior diaphragm, a defect in diaphragmatic pericardium and congenital intra-cardiac defects.[1,2]

Depending on the location of the protruding heart, and extent of the abdominal wall defect, ectopia cordis can be divided into cervical, cervico-thoracic, thoracic, thoraco-abdominal, and abdominal types. Tausing, in 1982 described the commonest form as the thoraco-abdominal type which typifies our index presentation.[4] The ectopic heart can be covered by omphalocele-like membrane or open (naked) in some instances.[5]

There are few cases of POC reported from Africa, mostly from Nigeria with poor treatment outcome.[6-9,10-12] To the best of our knowledge, only a few cases of Pentalogy of Cantrell have reported the problems associated with its management in a poor resource setting like ours.[2,8,12]

Full PC occurs rarely. Presence of the pentad is considered by some clinicians as full spectrum, with absence of some components seen as incomplete form of pentalogy which mirrors our case presentation.[13,14]

Cantrell's pentalogy is a very rare structural congenital syndrome with an estimated incidence put at 5.5 per 1 million live births[7] with very few cases reported in Nigeria.[8,9,11,12,14] POC is 2.7 times more common in boys, and African Americans may be more predisposed.[14]

Affected females usually present more severe symptoms,[15] which this index patient is also a female with incomplete Pentalogy of Cantrell, and the exact cause of Pentalogy of Cantrell has remained elusive. The cases are mostly sporadic, however many familial cases have been reported suggesting that genetic factors may play a role. It has been seen in chromosomal abnormalities like trisomy 21, trisomy 18, Turner syndrome and therefore, prenatal chromosomal studies are highly desirable.[13, 16-18]

Mutations implicated are deletions or duplication of the PORCN, thoraco-abdominal syndrome (TAS), teneurin-4 (TENM4), and ALDH1A2 genes.[19] The thoraco-abdominal syndrome including the pentalogy of Cantrell is suggested to be an X-linked dominant disorder, with mapped Xq24-q27 gene.[17] The mutagens linked to the POC include environmental, pharmacological, chemical, radiological et cetera.[20,21] In this case presented, there was no report of maternal exposure to any of these mutagens during early first trimester of pregnancy. The neonate also did not have dysmorphic features suggestive of a chromosomal disorder. There is no established association between maternal intrauterine infection and the development of Cantrell's pentalogy.[21] The pathogenesis of this syndrome has not been well elucidated, however one hypothesis that has enjoyed wide acceptance is that it results from a developmental anomaly. The error in embryogenesis appears to occur in a segment of the lateral mesoderm. The cardiac abnormalities are due to a faulty development of the epi-myocardium and the sternal and abdominal wall defects represent a faulty ventromedial migration of the paired mesodermal primordial structures. The diaphragmatic and pericardiac defects are secondary to a developmental failure of the septum transversum. These abnormalities seem to occur around day 14 and 18 of embryonic life.[1,14,22] Sowande and colleagues believe that the teratogenesis of this syndrome can be linked to band disruption following rupture of the chorion or yolk sac. The concept seeks to explain the numerous malformations seen in infants exclusive of torso.[14]

Tayoma in 1972 [3,14,15] after review of 61 cases of POC suggested a classification system:

i. Class 1, definite diagnosis with all five defects present.
ii. Class 2, probable diagnosis, with four defects including intracardiac and ventral abdominal wall abnormalities present.
iii. Class 3, incomplete expression, with various combination of defects, including sternal abnormality present.

Our patient may be regarded as incomplete expression of the syndrome as we were unable to demonstrate any intracardiac defect clinically and without an echocardiography as patient's care givers discharged against medical advice. Echocardiographic findings as documented in the literature shows these spectrums of anomalies in patients with cardiac defect; ASD (53%), Tetralogy of Fallot (20%) and ventricular diverticulum (20%) respectively.[1] This syndrome has variable expressivity with majority of those reported having the incomplete variant. Various other anomalies such as craniofacial like cleft lip and palate, and other CNS anomalies like encephalocele, hydrocephalus, cranio-rachischisis, and limb defects such as clubfoot, absent tibia, or radius and hypodactyly reported by other authors were absent in our index patient.[23-28]

Prenatal diagnosis of POC using obstetrics ultrasound scan at first trimester has developed significantly over the years since its first report in 1991.[28,29] This is thought to help in planning for appropriate management before delivery and provides information for parental counseling. Intrauterine diagnosis of this pentalogy is impossible before 12th week of gestation, because physiological bowel herniation occurs at this time.[30] Ultrasonography becomes a useful diagnostic method after this event.[22,31,32] However, our patient's anomalies were not detected in the antenatal scan carried out during the second trimester of pregnancy which could be due to it operator dependency.

The treatment of POC is tailored towards the presence and extent of any of the five defects. Reconstruction and repair can be single or multistage and requires both interdisciplinary and multidisciplinary team approach.[5,33] The survival rate of this syndrome is estimated to be around 37% and prognosis depends on the severity of intra-cardiac and extra-cardiac defects.[5] We had planned to evaluate the newborn holistically and repair the defect depending on severity but could not eventually carry that out as parents discharged the neonate against medical advice.

Over 71 million Nigerians are living in extreme poverty and a total of 133 million people classed as multidimensionally poor according to reports by world poverty clock 2023 and National Bureau of Statistics.[34] A paltry percentage (5.75%)[35] of Nigerian national budget is allocated to health in contrast to at least 15%

under the Abuja declaration with increasing catastrophic expenditure on the part of the care givers as a greater percentage of them access health care by paying out of pocket and this partly hindered further treatment of this vulnerable patient in our sub-region. Other worrisome problems in our sub-region are ignorance, high rate of illiteracy, superstitious beliefs, and cultural practices.[12, 36]

Policy makers regard paediatric surgery as being capital intensive, and catering for an insignificant minority, and hence pay less attention to providing health insurance coverage for the vulnerable paediatric patients.[37] With improvement in National Health Insurance Authority (NHIA) coverage, and increase access to paediatric surgical care, it could be a step in the right direction in obviating these occurrences of discharge against medical advice in our sub region.

CONCLUSION

Pentalogy of Cantrell is a rare congenital anomaly that requires a multidisciplinary management. The constraints we encountered in the initial management of this neonate is not uncommon in surgical practice within sub-Saharan Africa especially amongst carers of paediatric patients who live below the poverty line, with increasing catastrophic expenditure. We advocate health insurance (NHIA) coverage for a greater percentage of our vulnerable paediatric patients. This can be achieved by improving health care funding, and reducing the unmet surgical needs in our paediatric population. The preferred approach to management still needs to be established in resource constrained countries like ours.

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