Ectopia Cordis: Case Series and Literature Review

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Abstract

Ectopia cordis (EC) is a rare congenital malformation with partial or complete protrusion of the heart outside the thorax. It was first described by Haller in 1706. It can occur as an isolated lesion or as part of Cantrell's pentalogy. The thoraco-abdominal type of Ectopia cordis is usually associated with this distinct pentalogy. We reported 2 cases, the first was a 6-month-old female infant with partial ectopia cordis and suspected Pentalogy of Cantrell, who had survived with the condition without any medical or surgical intervention; but however presented with severe respiratory distress, cyanosis, and tachycardia which were in keeping with cardiac decompensation. The second was a late preterm neonate with thoracic type ectopia cordis diagnosed at birth, the baby was managed for problems of prematurity: respiratory distress syndrome, sepsis, jaundice, and anaemia, and was subsequently referred for surgery. We, therefore, draw the attention to Paediatricians and Obstetricians that early identification and management would most likely improve the survival of patients with ectopia cordis.

Keywords: Ectopia cordis, Cantrell, Pentalogy, Thoracoabdominal

I. INTRODUCTION

Ectopia cordis is a rare congenital cardiac anomaly associated with the partial or complete placement of the heart outside the thoracic cavity.[1] The term Ectopia is derived from the Greek word *ektopos*, meaning "away from."[1, 2] Based on the location of the heart, it can be classified into five types: cervical, cervicothoracic, thoracic, thoracoabdominal and abdominal. The first known case of Ectopia cordis was reported by Neil Stensen in 1671, it was however, first described by Haller in 1706.[1] It is usually of sporadic malformation, with reports associating it to chromosomal abnormalities like Turner syndrome, trisomy 18, 46 XX and 17q+ and a hallmark of Pentalogy of Cantrell.[3-6]

Although it remains rare, cases have been reported in Nigeria;[5,7] ectopia cordis affects all races equally with an estimated prevalence of 5.5 to 7.9 per million live births, with a slight male predominance.[1-3] It accounts for about 0.1% of cardiac anomalies.[8] The etiology remains unknown and diagnosis could be made prenatally with conventional ultrasound scan usually in the second trimester.[8-10]

Management is multidisciplinary and treatment usually involves staged surgical repair.[11] This is however hampered in our setting due to limited diagnostics, poverty and late presentation. We report these cases to highlight that though rare and having poor prognosis, the condition still occurs.

II. CASE 1

A 6-month-old female infant seen for the first time in our facility, on account of difficulty in breathing and fast breathing of about 9 hours duration. There was no history of cough, or any history suggestive of force feeding. The child sucks from the breast directly. The child had a swelling, just below the anterior chest wall extending to the upper anterior abdominal wall, with a healed scar extending towards the umbilicus. The swelling was pulsatile and covered by a thin membrane. It was said to have progressively reduced in size since birth. Pregnancy was not adversely eventful, no exposure to teratogen substances or use of herbal preparations. Pregnancy was carried to term and delivery was at a private hospital, APGAR score and birth weight could not be ascertained.

There was no immediate postnatal period admission for sepsis or neonatal jaundice. On examination, child was acutely ill looking in severe respiratory distress, cyanosed, febrile (T – 37.80), not pale, anicteric, not dehydrated, and no finger clubbing. There was a swelling located mainly in the epigastric region, having a thin membranous covering, pulsatile, measured about 6 x 8cm (figure 1).

She was tachypneic with respiratory rate of 68c/m, oxygen saturation of 60% in room air, and tachycardic (heart rate – 186b/m) with heart sound S1, S2 and systolic murmur heard on the swelling. She was placed in cardiac position with the knee to the chest, which improved the cyanosis. Intranasal oxygen was commenced. Samples were taken for full blood count and serum electrolytes, urea and creatinine. Chest radiograph, Electrocardiography and Echocardiography were ordered. However, few minutes into admission, child suddenly became apneic and all resuscitative measures proved abortive. Postmortem could not be done because the parents declined.

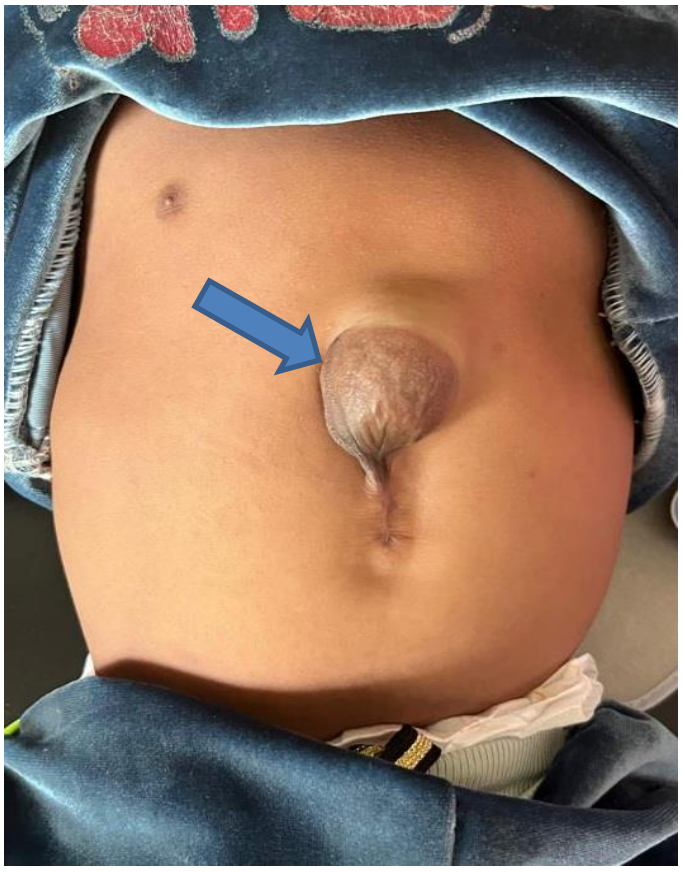

FIGURE 1: Shows the protruding heart covered with a thin skin membrane

III. CASE 2

A late preterm (36 weeks) female neonate delivered in our facility via spontaneous vaginal delivery. The mother had regular antenatal care visits, haematinics and received 2 doses of tetanus toxoid and malaria prophylaxis. There was no premature rupture of membrane (PROM) or dysuria. She was delivered via spontaneous vaginal delivery (SVD), baby had an APGAR score of 6 and 7 in the 1st and 5th minutes respectively and a birth weight of 2.5kg. The oxygen saturation was 98%. There was an obvious chest wall deformity with visible pulsation. She also had a whitish opacity in the right eye.

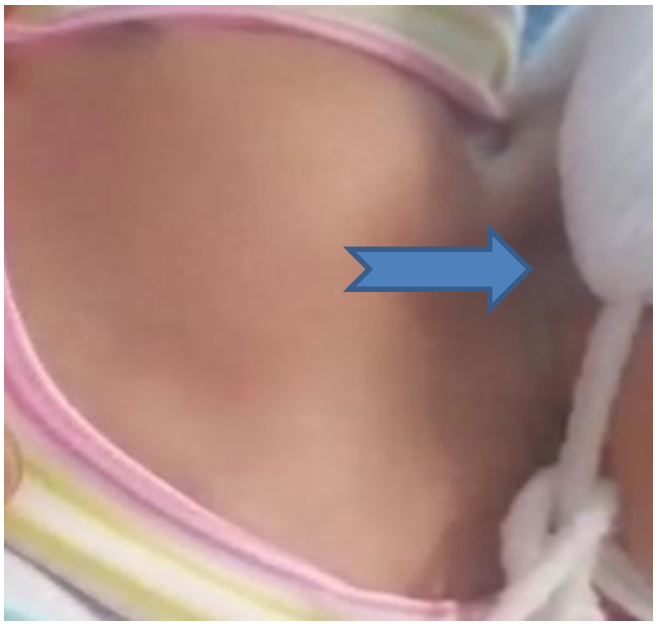

FIGURE 2: Pre-surgery showing depression of the upper anterior chest wall defect

The baby's vitals and examination findings were within normal for expected, however, she was admitted for anticipatory care of the preterm with congenital chest wall defect. At the third day of life, she was noticed to be anaemic and icteric, packed cell volume was 35%, which necessitated transfusion. She was also commenced on phototherapy. She had chest X-ray done, which showed wide apart medial sternal clavicular ends, globular heart shape with elevated apex and lungs appeared plethoric.

Chest ultrasound scan showed midline anterior thoracic bony wall defect, heart seen in the midline covered anteriorly only with skin, subcutaneous tissue and muscle, not protruding beyond the chest wall. Thickened right ventricular wall, measured 7.8mm when compared to the left which measured 3.8mm. Electrocardiography showed right axis deviation possibly an old inferior myocardial infarction.

A diagnosis of partial ectopic cordis was made, the parents were counselled on the diagnosis, line of

management and prognosis. She received antibiotics, fluids, oxygen, phototherapy, and supplements, and was discharged after spending 11 days on admission. She was referred to another teaching hospital where the chest defect was closed by a cardio thoracic surgeon. She is attending a clinic visit with the Neonatologist and Ophthalmologist.

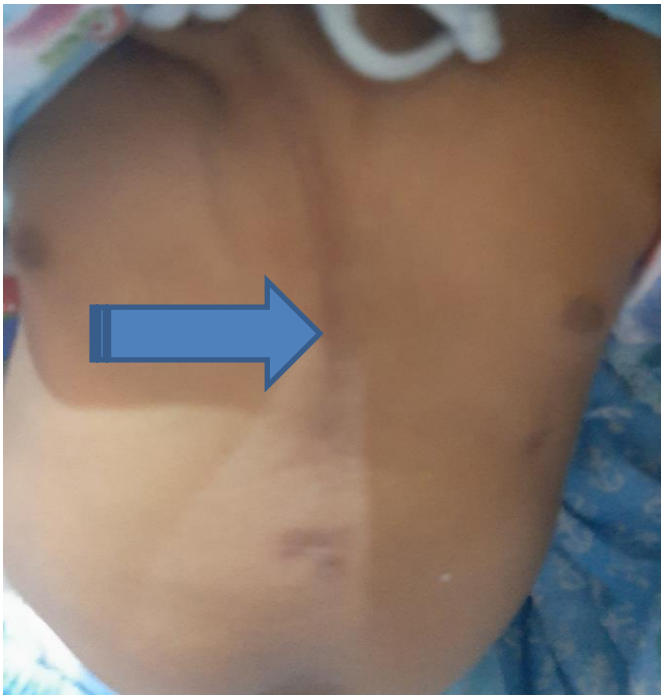

FIGURE 3: Shows post-surgical scar following closure of the anterior chest wall defect.

IV. DISCUSSION

The failure of the paired cartilaginous bar of the embryonic sternum to fuse leads to sternal cleft. This may lead to the partial or complete location of the heart outside the thorax, known as Ectopia cordis. Weese in 1818 and Todd in 1836, classified ectopia cordis into five different types: cervical, cervicothoracic, thoracic, thoracoabdominal, or abdominal.[1, 12] Byron, however classified it into four different types, with the exception of the cervicothoracic type. The most common types are thoracoabdominal and abdominal. The prognosis depends on the clinical type of ectopia cordis.[5, 10]

The abdominal type has a favorable prognosis while the cervical and thoracic have the worst prognosis.[13] The thoracoabdominal type is associated with another rare and distinct syndrome known as Cantrell´s pentalogy, first described in 1958 by Cantrell et al.[14] The pentad consists of: distal sternum defect; midline supra-umbilical abdominal wall defect; ventral diaphragmatic hernia; defect of the anterior diaphragmatic pericardium; and congenital intracardiac defects such as ventricular septal defect, atrial septal defect and tetralogy of fallot.[14,15]

The presentation of the first patient was consistent with the thoracoabdominal type of ectopia cordis, with associated distal sternum defect and a midline supra-umbilical abdominal wall defect, the other component of the pentad could not be investigated before the child succumbed to the condition. Autopsy could have been helpful in highlighting the intracardiac defects, however parents declined. It could be inferred that there is a possibility of tetralogy of fallot like heart lesion because the cyanosis in the child reduced when she was placed in knee chest position. Ectopia cordis is associated with both intracardiac and extracardiac lesions.[8] Ugowe *et al*,[5] reported a case in Ife, in 2019, a 7-day old preterm male neonate having ectopia cordis with multiple cardiac defects, sternal and abdominal wall defect, diaphragmatic and pericardial wall defects seen on ultrasound and echocardiography respectively.

In Kano, Mukhtar-Yola *et al*[7] also noted similar findings in a term neonate. Similarly, Aliyu and Mohammed were able to demonstrate the complete expression of the Cantrell's pantalogy in a nine-month-old boy in Kano, Nigeria.[15] Other variants were reported in Akure,[6] Maiduguri,[3] and Cameroon.[11] The different clinical features of the cases reported could be due to the time of presentation, associated comorbidities, investigations done, and interventions provided. Pentalogy of Cantrell is associated with high mortality, the survival rate is lower than 40%, and the prognosis depends on the severity of cardiac and extracardiac anomalies.[10]

The second patient with the thoracic type ectopia cordis had surgical closure and was on follow up post-surgery. In a cohort study[8] of 31 cases, all survivors were those who were born alive and underwent surgery. Ectopic cordis can be diagnosed prenatally in the first trimester and during specialized anomaly scanning in the second trimester. In the cases we reported, there was no evidence of the ectopia cordis detected during prenatal ultrasound scan. This will allow for adequate parental preparation and multidisciplinary care can be planned before delivery.

V. CONCLUSION

We have presented two cases of Ectopia cordis; - a thoracic type and a thoracoabdominal type. While the latter case has a better prognosis, this can become

worse when associated with congenital anomalies and late presentation as reported in our case. The thoracic type reported had a favourable outcome due to prompt diagnosis and surgical intervention. Prenatal diagnosis using ultrasonography is currently of great value. A multidisciplinary approach is necessary to limit morbidity and mortality when the diagnosis is made.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

AUTHORS' CONTRIBUTIONS

ABF and AD conceptualized the study and wrote the initial draft of manuscript; AD wrote the literature review. ABF, AD, AJO and ABJ reviewed the manuscript, and made significant contributions. All authors read and approved the final manuscript

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