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ECTOPIA CORDIS: REPORT OF TWO CASES

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SUMMARY

Ectopia Cordis is a dramatic but rare cardiac anomaly with poor prognosis in most centers worldwide. This rare malformation occurs in 5.5-7.9 per million live births in the USA(1). This cardiac malformation is defined as a heart that is in an extra- thoracic position. In this article, we report two newborn infants admitted to our newborn unit with the heart beating outside the thoracic cavity. One child succumbed due to prematurity and severe cardiac malformations, while the other child successfully underwent surgical reconstruction of the ectopia cordis. She succumbed due to overwhelming sepsis one week after the surgery. Such cases have neither been reported nor treated locally.

INTRODUCTION

Ectopia cordis represents a form of pericardial defect that is further associated with displacement of the heart outside the thoracic cavity. There are various frequently recognised positions of the heart in ectopia cordis. Van Praagh *et al* classified ectopia cordis as represented by four types: cervical, thoracic, thoraco-abdominal and abdominal(2). The anatomy of the heart may be normal but structural cardiac defects and other non-cardiac malformations commonly accompany the condition and impact negatively on prognosis. Aetiology of the condition is unknown.

CASE REPORTS

Case 1: G.N, a male infant delivered at the Pumwani Maternity Hospital in Nairobi by spontaneous vertex delivery and transferred to the Kenyatta National Referral and Teaching Hospital the same day on 9th December 2003. He had low apgar scores of 4:1 and 6:5 min and weighed 2kg. The mother had used injectable contraceptive "depo provera" for two years before conception. She never attended any antenatal clinic, had no antenatal profile and only went to hospital when she went into spontaneous labour. Her last menstrual period was on 28th April 2003 and the expected date of delivery was 4th February 2004. Gestation at birth was thirty-two weeks by dates.

On physical examination, the child looked dull and lethargic with a weak cry. He had no pallor, or jaundice but had acrocyanosis with mild pedal edema. The pulses were regular and of good volume and there was no radio-femoral delay. There was a mid sternal cleft from where the heart protruded, actively pulsating with the apex pointing upwards. Both atrial appendages were visible. The origins of the great arteries dipped into the thoracic cavity. The pericardium was absent and instead there were some greenish material covering the heart (Figure 1). The umbilical cord arose below the heart and was normal. The first and second heart sounds were normal and there were no murmurs. Chest auscultation was normal. The liver and spleen were not palpable.

A two dimensional echocardiogram done immediately showed situs solitus with mesocardia, atrioventricular and ventriculoarterial concordance. The atrial septum was intact but there was a very large perimembranous inlet /outlet ventricular septal defect with aortic override. No pulmonary artery stenosis. The ductus had closed. Haemogram showed haemoglobin of 16.7gm/dl, the total white cell count of 16.3x 10/9, with differential count of 88% polymorphs and 10% lymphocytes, the platelets were adequate.

In view of the poor general condition and the severe cardiac malformation, it was decided to manage baby G.N conservatively. The heart was covered with sterile gauze that was constantly irrigated with warm normal saline, incubator care and feeding through nasogastric tube was started. Intravenous antibiotics crystalline penicillin and gentamycin were commenced for presumed sepsis. At around 3 am on 10th February 2004, the baby developed progressive bradycardia. No resuscitation was given and the baby succumbed one hour later. The parents declined consent for postmortem studies.

Case 2: The second baby A.M was a female infant born on 20th September 2003 at home to a 20-year-old Para 2+0, gravida 3 mother, following an uneventful

pregnancy. She cried immediately at birth. The mother had attended antenatal clinic from seven months gestation, the antenatal profile was normal although she never had any obstetric ultrasonography. She had used oral contraceptive pills up to six months before conception. The membranes ruptured during labour and she was delivered by spontaneous vertex delivery. The mother said the baby cried immediately. Her two other children both females aged three and five years are alive and well.

At admission, this was a female baby of good body colour, with acrocyanosis. The body weight was 3000gms. She was not pale, had no edema or jaundice. The main malformation was at the chest whereby the sternum was missing with the lower portion of the chest devoid of skin but the upper part had thin hyper-pigmented skin. The heart protruded just at the level of what should have been the xiphoid process. It was actively pulsatile with no membrane cover. The ventricle (probably left) and part of the atrial appendage were exposed; the great vessels were buried at the level of the sternal angle. The upper abdominal wall was deficient (omphalocele) with the umbilicus in an elevated position, the cardiac apex was not uplifted. The diaphragm was exposed (Figure 2). The umbilicus had two arteries and one vein. The pulses were regular, good volume with no radio femoral delay. The first and second heart sounds were normal with no murmurs. The breath sounds were normal and there were no added sounds on chest auscultation. Central nervous system was normal. There was no cleft lip or palate. A diagnosis was made of ectopia cordis to rule out any intracardiac anomalies.

Figure 1

Picture of baby G.N. Showing sternal cleft with protrusion of the heart outside the chest cavity and cardiac apex pointing upwards

Figure 2

Preoperative picture of baby A.M. showing exposed heart, diaphragm and umbilical cord. The cardiac apex is downward and to the left

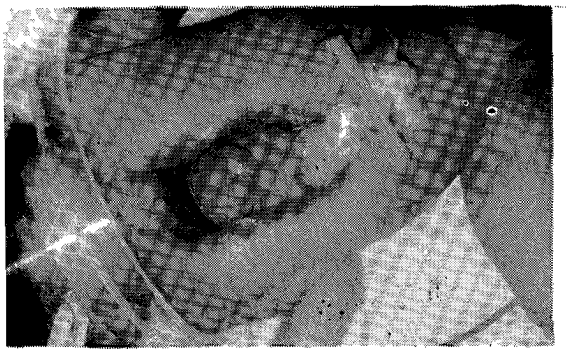
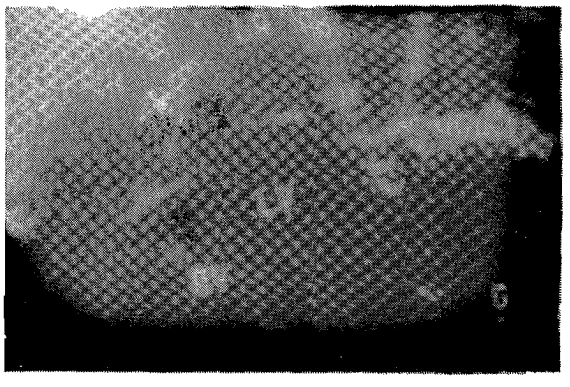


Figure 3

Postoperative picture of baby A.M. showing repair of the abdomen and repositioning of the heart in the thoracic cavity

Figure 4

Postoperative echocardiogram an apical four chamber view of baby A.M. showing an elongated left and right ventricle



Upon admission, the baby was commenced on 10% dextrose, with supplemental sodium and potassium and nursed in an incubator. Continuous electrocardiograms and percutaneous oxygen saturations were monitored. Supplemental oxygen (100%) was given by oxyhood at 5L/min. The heart and the exposed omphalocele were covered with sterile gauze moistened with normal saline four hourly. Investigations done included full haemograms, urea and electrolytes, blood sugar and chest X-ray. Consultations of various specialists including paediatric cardiologists, cardiothoracic surgeons, plastic surgeons, anaesthetists and the intensive care personnel was done, and the patient was started on prophylactic antibiotics (cefuroxime 50mg/kg/day three times per day). The baby remained stable with percutaneous saturations ranging from 96 to 100% on oxygen and between 90 to 96% off oxygen. The haemoglobin was 17.2gm/dl, random blood sugar, urea and electrolytes were normal. A chest X-ray showed complete absence of the sternum, normal pulmonary vascular markings and a midline chest deformity. A two dimensional echocardiogram and colour flow map showed situs solitus with a levocardia. Two atria and two ventricles were seen with a patent foramen ovale. The ventricular septum was intact. The great vessel relationship was normal and there was good kinesis of the left ventricle. The electrocardiogram tracing on the monitor had a heart rate that varied between 130 to 140 beats per minute with a normal sinus rhythm and normal P waves. Both parents were counseled and the patient prepared for theatre with the plastic surgeon and cardiothoracic surgeons as the team leaders. Surgery was done on day 2.

At surgery, the plastic surgeons successfully raised a fascia- cutaneous flap to cover the exposed heart and abdomen (Figure 3). The patient withstood surgery well and she was subsequently transferred to the intensive care unit. In the intensive care unit, the baby maintained good oxygen saturations. Enteral feeds of expressed breast milk 28mls 4hrly was started through nasogastric tube. An attempt to wean the baby from the ventilator was deferred initially due to severe chest indrawings and later due to "systemic desaturations". A repeat echocardiogram showed an elongated left ventricle with systolic obliteration of the mid portion. The diastolic function was normal (Figure 4).

On the fifth day, the baby developed jaundice that rapidly progressed and the haemoglobin fell to 13.9gm/dl. After taking blood for bilirubin levels, phototherapy was commenced and a diagnosis of sepsis considered. A septic screen excluding lumber puncture was ordered followed by an empirical change of antibiotics to second line amikacin and ceftriaxone, she continued to tolerate feeds, which were, increased to 34mls four hourly.

On the sixth day the baby developed sclerema, the jaundice deepened and her saturations started to fall. She had episodes of hypothermia associated with relative bradycardia (116-120, down from 140 beats per minute), this was followed by abdominal distension so enteral feeds were stopped. Her total neutrophil count increased to $18.9x10^9$ with thrombocytopenia. The baby succumbed to overwhelming sepsis on the sixth postoperative day. Blood culture report received after death grew pseudomonas.

DISCUSSION

Ectopia cordis being a rare condition, has taken time to evolve a rational approach to its management. Management principally is surgical involving staged interventions to; (a) Cover the naked heart, b) Placement of the heart into the thoracic cavity, c) Sternal and or thoracic cage reconstruction(3-5). In the developing countries, apart from the known high mortality, one has to contend with limited facilities to carry out staged surgery and lack of adequate intensive care facilities(6).

The causation of the condition has not been identified; Steichert *et al* in the Czech Republic have been able to induce ectopia cordis in chick embryos of 84% of cases subjected to intra - amniotic injection of hydrocortisone. The relevance of this to humans needs to be studied(7). Of the various types of ectopia cordis, the cervical type is rare and is observed with sternum intact. It represents the retention of the heart in its embryonic position in the neck. The thoracic type is the classical type of ectopia cordis. It is characterised by a sternal cleft that allows the protrusion of the heart outside the chest cavity and is associated with cephalic orientation of the cardiac apex, epigastric omphalocele, and small thoracic cavity. This form of ectopia cordis has been associated with congenital heart diseases like tetralogy of fallot, ventricular hypoplasia, transposition of the great arteries and double outlet right ventricle. We believe our first baby G.N. had this form of ectopia cordis. Other extra cardiac anomalies reported in thoracic type include meningocele, prolapse of the forebrain, cleft lip and palate(8). None of these were observed in the two cases.

The thoraco-abdominal type represents a constellation of associated anomalies sometimes referred to as pentalogy of cantrell. These comprise midline supraumbilical abdominal defect, defect of distal sternum, deficiency of diaphragmatic pericardium, deficiency of anterior diaphragm and congenital cardiac anomalies(9). We believe our second patient (A.M) had the thoraco-abdominal form of ectopia cordis with total lack of the sternum. She did not have the cephalic orientation that is commonly described in this series(9,10), and was lucky not to have any additional intra-cardiac or extra-cardiac congenital malformations.

The abdominal type is extremely rare and appears to represent a diaphragmatic defect with continued migration of the heart into the abdominal cavity. Baby A.M withstood the surgery quite well, on the second postoperative day she was weaned off the ventilator; however there was severe chest retractions associated

with arterial oxygen desaturations hence the baby had to be put back on mechanical ventilation. Several factors thought to have contributed to the overwhelming sepsis in this patient included; i) Home delivery, and subsequent transportation of the baby to the hospital under questionable hygienic conditions, ii) The delay in mobilization of the team making surgical intervention to be performed on the second day of birth, iii) This peculiar malformation seen in this hospital for the first time also attracted curiosity among hospital workers making the risk of added nosocomial infections even greater.

An echocardiogram done on the fourth postoperative day showed markedly elongated left ventricle with systolic obliteration of the apical segments could have contributed to unforeseen ventricular dysfunctions and the subsequent difficulties in weaning the patient off the ventilator. Most patients with ectopia cordis present incidentally after birth, but prenatal diagnosis on ultrasound is now possible in centers with experience in foetal echocardiography and ultrasonography(7,11,14). Despite surgical intervention, some centers in the developed world still report 100% mortality (11,15).

The surgical management of thoracic ectopia cordis involves initial internalization of the heart by bilateral pectoral skin flaps followed fourteen months later by a definitive repair to reconstruct the anterior chest wall with prosthesis(3,4,6,12,13). This can sometimes be associated with kinking of the great vessels especially when there is associated cephalic orientation of the cardiac apex. In the case of A.M, this was not thought to have contributed to the mortality. While the first stage of repair was largely successful, the cause of death was overwhelming septicaemia in a situation where there was no specialized neonatal intensive care.

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