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CASE REPORT

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Pentalogy of Cantrell - A Case Report from Nigeria.

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Abstract: FB was delivered at home to a para three low income mother, at term after an unsupervised pregnancy. At birth she was noticed to have an anterior chest wall defect with a protruding pulsating mass. Further examination revealed a defect from the suprasternal notch to the xiphoid process (bifid sternum) with the heart exposed and pulsating (ectopia cordis). The pericardium was absent and the great vessels were exposed. There was an epigastric omphalocoele and a ventral diaphragmatic defect. The diagnosis of Pentallogy of Cantrell was made. The challenges involved in the evaluation and management of this case in a Nigerian setting are discussed.

Keywords: Pentallogy, Cantrell, Nigeria.

Introduction

The pentalogy of Cantrell is a rare syndrome with an estimated incidence of 5.5 per 1 million live birth [1] This syndrome was first described by Cantrell et all [2] in 1958 to include a pentad of findings of a midline supra umbilical thoracoabdominal wall defect, a defect in the lower sternum, a deficiency of the diaphragmatic pericardium, a deficiency of the anterior diaphragm and various congenital cardiac abnormalities including an ectopia cordis. The Ectopia cordis may be complete or partial. The pathogenesis of pentalogy of Cantrell has not been fully elucidated. Cantrell et al. [2] suggested an embryologic developmental failure of a segment of the lateral mesoderm around gestational age 1418 days. Consequently, the transverse septum of the diaphragm does not develop, and the paired mesodermal folds of the upper abdomen do not migrate ventromedially. Organs may eviscerate through the resulting sternal and abdominal wall defects.

The pentalogy is said to be fully expressed when all the five components are present, however other variants have been described by Toyoma et al.[3] He suggested the following classification of the pentalogy of Cantrell: class 1, definite diagnosis, with all five defects present; class 2, probable diagnosis, with four defects present, including intracardiac and ventral wall abnormalities; and class 3, incomplete expression, with various combinations of defects present, including a sternal abnormality. A review of the literature yielded 58 reported patients with pentalogy of Cantrell between 1987 and April 2007.[4] Thirty-three patients were described as complete and 23 patients as incomplete. Two patients were not clearly defined as complete or incomplete.

The prognosis in these patients depends on the severity of the cardiac anomalies and is usually very poor, however a few cases have been succesfully operated on. There is a paucity of local data from Africa and to our knowledge this is the first case being reported from Nigeria in Africa.

Case summary

Baby FB was referred to the special care baby unit (SCBU) of Aminu Kano Teaching Hospital Kano (AKTH) at the age of 4 hours. She was a product of term gestation delivered to a 23 year old Para 3 (all alive and well) non diabetic non hypertensive mother. Pregnancy was uneventful; mother had 2 visits to a primary health care centre but delivered at home as she did with her other children. Ultrasound scan was not done in pregnancy. Baby cried well at birth and was immediately noticed to have an anterior chest wall defect with a protruding pulsating mass, and she was transferred to AKTH. There was no history of congenital malformation in the family, mother is a full time house wife and father is a petty trader, both are educated to primary school level and are low income earners.

Examination revealed an active, uniformly pink, anicteric, acyanosed baby drooling saliva. She weighed 3.2kg with a length of 50cm and occipitofrontal circumference of 35cm which are all appropriate for gestational age. There were no dysmorphic facies, head and neck were normal. The anterior chest wall revealed a defect from the suprasternal notch to the xiphoid process (complete bifid sternum) with the heart exposed and pulsating supported only by the great vessels (complete ectopia cordis). The pericardium was absent and the great vessels were exposed. There was an epigastric omphalocoele and a ventral diaphragmatic defect. Other systems were essentially normal. The diagnosis of Pentallogy of Cantrell was made.

Baby was admitted and barrier nursed. Complete blood count, urea and electrolytes, urinalysis, random blood sugar were all within normal limits. The chest X ray (CXR)is as shown in fig.1, Transesophageal/Doppler echocardiography, video oesophagogram and blood gases /oxygen saturation could not be done as the facilities were unavailable. So it was not possible to definately determine presence of intracardiac abnormalities.

She was commenced on oxygen, she was suctioned PRN and commenced on 10% dextrose water, Vitamin K and IV ceftazidime and gentamicin. Strict input and output chart was maintained and the chest wall defect was covered with sofratulle.

A multidisciplinary team involving the paediatrician, paediatric and vascular surgeons, anaesthetist, cardiologist, social welfare was constituted. Parents were counseled on the pros and cons for a temporizing surgery prior to transfer to a cardiothoracic surgical centre abroad if a government waiver was obtained (This normaly takes several days or weeks). Surgery was done on the 2 nd day of life with the aim of mobilizing skin to temporarily cover the heart before transport. The intra operative findings were those of an exposed heart, epigastric omphalocele, liver dome in chest, complete bifid sternum and suggestive of a left to right shunt.

Baby was admitted in the intensive care unit, however 3 hours after surgery her condition deteriorated following cardiac arrest, she was resuscitated, ventilated, stitches released but all efforts proved abortive and she died at the age of 39 hours. The parents did not consent to a post mortem examination.

Fig.1

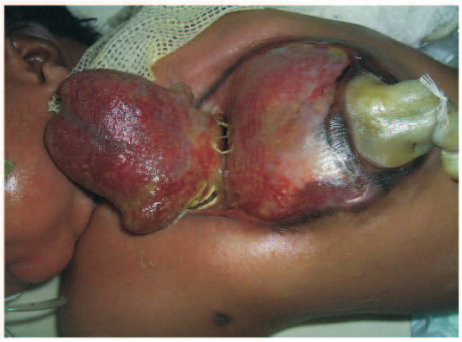


Fig.2

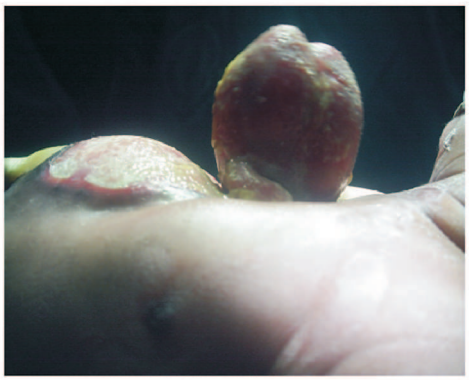


Fig.3

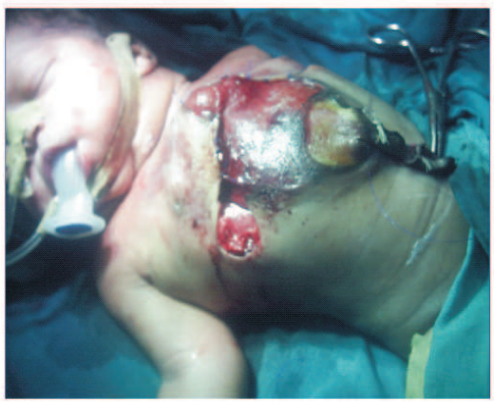
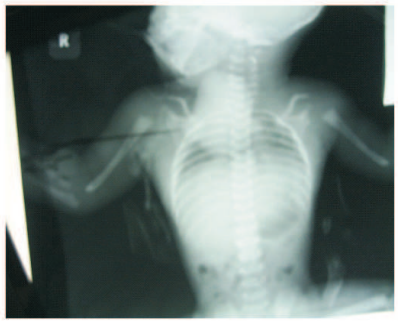


Fig 4



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Discussion

The pentalogy of Cantrell is estimated to have an incidence of 1:65,000 live births [5.] The main abdominal wall malformation associated with this syndrome is omphalocele found in 74.5% of patients followed by deformed regions in the lower sternum (59.4%), diaphragm (56.8%), and pericardium (41.8%). Cardiac anomalies occurs in 83% of cases with interventricular communications as the most common intracardiac abnormality. Intra-cardiac anomalies described include ventricular septal defect (100%), atrial septal defect (53%), tetralogy of Fallot (20%), and ventricular diverticulum (20%). [2]

Our patient had an extrathoracic ectopia cordis but we could not determine if he had an associated intra cardiac lesion as we could not do a transoesophageal echocardiography. In the most common thoracic forms of ectopia cordis, the sternum is split and the heart protrudes outside the chest. In other forms, the heart protrudes through the diaphraghm into the abdominal cavity or may be situated in the neck. [6] Associated anomalies have been reported with POC and include craniofacial and central nervous system anomalies such as cleft lip and/or palate, encephalocele, hydrocephalus, craniorachischisis [7,8,9] limb defects such as clubfoot, absence of tibia or radius, and hypodactyly [10,11] also reported are abdominal organ defects such as gallblader agenesis and polysplenia .[12]

With prenatal ultrasonography, POC usually can be diagnosed in the first trimester of pregnancy. [13] Use of prenatal magnetic resonance imaging (MRI) may enhance the visualization of the fetal anomalies and help in planning of surgeries. [14] In our patient the mother did not have adequate antenatal care and did not get an ultrasound done in pregnancy as such the diagnosis at birth was such a shock for the family and a great challenge for the managing doctors. We had a lot of challenges in managing this case, from investigations to management. This family is a low income family and did not have any form of insurance. So all health expenses were out of pocket spending and they had severe constrains. Though we had requested that the government should assist with the finances, we also knew that it will take a while before all the procedures could be completed and patient transfered abroad. Moreover we could not do some investigations because of lack of equipments. There is also no established cardiac centre in the country that we could immediately transfer the patient to. So in order to curb infection the multrtidisciplinary team agreed on a temorizing surgery prior to transfer.

Controversies exist with regards to the best surgical technique for this rare syndrome necessitating multiple operations. [5] The treatment of the POC consists of corrective or palliative cardiovascular surgery, correction of ventral hernia and diaphragmatic defects and correction of associated anomalies. The best treatment strategy depends on the size of the abdominal wall defect, the associated heart anomalies, and the type of EC. In most cases, death occurs in the first days of life, usually from infection, cardiac failure or hypoxaemia. [6] Surgical therapy for neonates without overwhelmingly severe cardiac abnormalities consists of covering the heart with skin without compromising venous return or ventricular ejection, palliation of associated defects is also very necessary. Occasional patients with the abdominal type of EC have survived to adult hood. [6]

Clinical parameters such as birth weight and immaturity, associated congenital heart anomalies, and size of the abdominal defects are the main elements to predict the outcome of these patients: particularly the prognosis of POC depends on the severity of the associated cardiac anomaly. The prognosis seems to be poorer in patients with the complete form of pentalogy of Cantrell, EC, and patients with associated anomalies.

African children face a great challenge as facilities to diagnose and care for complex problems such as this, are non- existant. When the diagnosis of pentalogy of Cantrell is suspected, a multidisciplinary approach is essential. A prenatal medical team consisting of a gynecologist, a neonatologist, pediatric cardiologist, pediatric surgeon and geneticist, should use their expertise in choosing the best approach to this severe disorder.

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