Pentalogy of Cantrell - A Case Report from Nigeria.

Introduction

The pentalogy of Cantrell is a rare syndrome with an estimated incidence of 5.5 per 1 million live birth. This syndrome was first described by Cantrell et all 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. 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.

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 Pentalogy of Cantrell was made. The challenges involved in the evaluation and management of this case in a Nigerian setting are discussed.

Keywords: Pentalogy, Cantrell, Nigeria.
**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 omphalocele and a ventral diaphragmatic defect. Other systems were essentially normal. The diagnosis of Pentalogy 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 normally takes several days or weeks). Surgery was done on the 2nd 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.

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![Fig.1](image1)

![Fig.2](image2)

![Fig.3](image3)
Discussion

The pentalogy of Cantrell is estimated to have an incidence of 1:65,000 live births. 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 intraventricular 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%).

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 diaphragm into the abdominal cavity or may be situated in the neck. 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 limb defects such as clubfoot, absence of tibia or radius, and hypodactyly also reported are abdominal organ defects such as gallblader agenesis and polysplenia.

With prenatal ultrasonography, POC usually can be diagnosed in the first trimester of pregnancy. Use of prenatal magnetic resonance imaging (MRI) may enhance the visualization of the fetal anomalies and help in planning of surgeries. 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 multidisciplinary team agreed on a temorizing surgery prior to transfer.

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References


