Pentalogy of Cantrell: A Report of Three Cases

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ABSTRACT

Pentalogy of Cantrell is a rare upper midline syndrome that may present in association with anomalies outside the torso. The pentad — the supraumbilical body wall defect, sternal defect, deficiency of the anterior diaphragm, defect of the diaphragmatic pericardium, and the intracardiac anomalies — was first described by Cantrell *et al*., in 1958. The defect is said to be more common in males, and survival is dependent on the cardiac malformations and on the degree of completeness of the syndrome. We report three cases of Cantrell's pentalogy managed in our unit. Two of the patients were females and one a male. All were seen at peripheral health centers before being referred to us. Age at presentation for the girls was 18 hours and 36 hours, respectively, the boy presented at the age of six weeks. All of their parents were unschooled manual workers. All patients presented with a defect in the supraumbilical body wall, bifid sternum, and a visible cardiac impulse. We were unable to do echocardiography to rule out intracardiac anomalies in the three patients. The thin membranous covering of the epigastrium in the female patients was managed conservatively. Both female patients were discharged against medical advice as requested by their parents, due to financial constraints. The male patient was lost to follow up after two clinic visits. A multidisciplinary approach to the management of this syndrome is recommended.

Key word: Pattern, pentalogy of cantrell, sternal defect

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INTRODUCTION

Cantrell *et al*.[1] reported five cases in 1958, describing a pentad of findings that included: (1) supraumbilical wall defect, (2) defect of the lower sternum, (3) deficiency of the anterior diaphragm, (4) defect of the diaphragmatic pericardium, and (5) various intracardiac defects. The pathogenesis of this syndrome is not fully understood. Occurrence of the full pentalogy is rare and the survival rate of the patients with complete pentalogy is low,[2] with outcome being dependent on the severity of the cardiac malformation, extra cardiac defects, and other associated anomalies. We report three cases of pentalogy of Cantrell seen in Ile – Ife, Nigeria, and our constraints in their management.

CASE REPORTS

Case 1

A female baby weighing 2.45 kg was born to a 20-year-old woman. The parents were not related. The child was the second for the parents; the first had died at one year of age from ill health. The baby was born in a peripheral health center and was 18 hours old when she was transferred to our hospital. Pregnancy and delivery were supervised and there was no history of maternal illness during the pregnancy. There was no history of a prenatal ultrasound diagnosis of the anomaly. There was no history suggestive of exposure to teratogenes during pregnancy. There was no family history of birth defects. The child cried spontaneously at birth. Examination revealed a non-cyanosed anicteric baby, who was not in any obvious respiratory distress. The chest examination showed an obvious anterior chest wall defect covered by skin, which was pink and membranous over the epigastrium. The cardiac impulse was visible. Palpation of the chest wall revealed a complete sternal defect. Breath sounds were well heard all over the chest, only the first and second heart sounds were heard over the precordium. The heart rate was 140 per minute. The umbilical stump was intact with no obvious anomaly of the umbilicus. No other anomalies were seen on examination [Figure 1].

Echocardiography could not be performed in our center because the echocardiography machine could not be adapted for pediatric use, and the parents could not afford the investigation in a private diagnostic center. The child was on admission for seven weeks, and 70% alcohol (ethanol) dressing of the membranous covering over the epigastrium was done daily to encourage epithelialization. The child remained clinically stable throughout the period of admission. The father who was a carpenter could not afford any further investigations, or the cost of admission, and requested for child's discharge against medical advice. Patient was lost to follow up.

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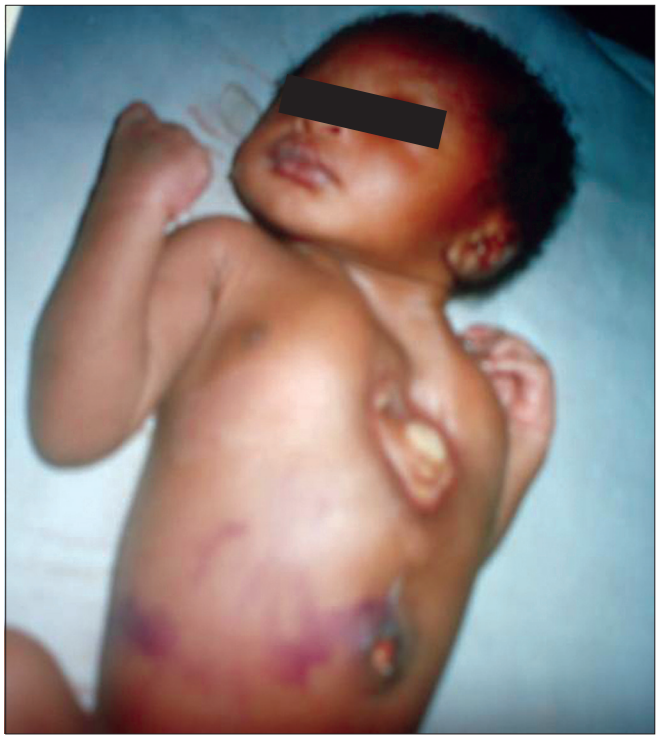

Figure 1: Child with epigastric omphalocoele

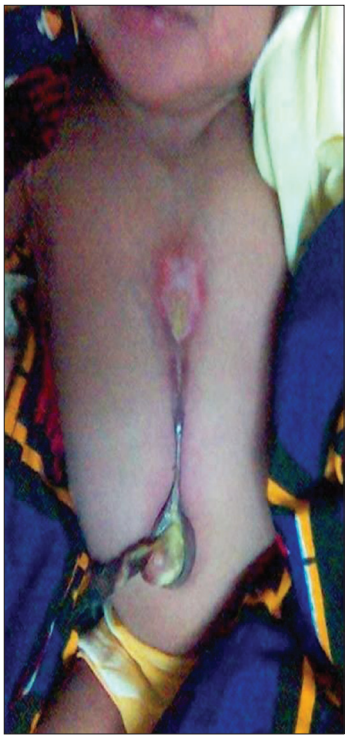

Figure 2: Noticeable midline fibrous raphe

Case 2

A 36-hour-old female child was brought to our hospital following referral from a health center. The child was born at home. Delivery was supervised by a birth attendant. The child cried spontaneously at birth, and the weight on presentation was 2.5 kg. The mother was a 30-year-old Para 3 (two were alive) woman. The first child died on the second day of life from unknown causes. There was no history of consanguinity among the parents. There was no history suggestive of exposure to teratogenes during pregnancy. Physical examination showed an acyanosed, anicteric child who had a right subconjuctival hemorrhage and was not in any obvious respiratory distress. A complete sternal cleft was noted on chest examination. Breath sounds were normal bilaterally, and the heart rate was 140 per minute. Only heart sounds I and II were heard. A thin membranous covering of the epigastrium about 3 cm in diameter was noted, with a raised fibrous raphe, of about 5 mm extending from the membranous covering of the epigastrium to a normal appearing umbilical stump [Figure 2]. The cardiac impulse was visible through the epigastric covering. No limb nor vertebral defects were noted. The anterior fontanelle was normotensive and head circumference was 33 cm.

A chest radiograph revealed a centrally placed globular heart, with good lung expansion and markings, bilaterally [Figure 3]. Echocardiography could not be done in our center, and the parents could not afford the investigation in a private diagnostic center. Daily dressing of the epigastric membranous covering was done with 70% alcohol. Parents who were peasants requested for the child's discharge, against medical advice, on financial grounds. The child was discharged on the sixth day of admission and has been lost to follow up.

Case 3

This patient was referred to our unit from a peripheral health center at six weeks of age. The child was referred on account of a chest wall defect, which was noticed at birth. The patient was the first child of the mother's second marriage. The mother was a remarried widow who had three other children who were alive and well. The father was a farmer. No history of difficulty in breathing. No history of cyanosis. Examination showed the child was not pale and had no obvious respiratory distress. Chest wall examination revealed a complete sternal defect with visible cardiac impulse beneath the anterior chest wall skin covering. The heart rate was 144 per minute, and only heart sounds I and II were heard, no murmur. The abdomen was flat and moved with respiration. The umbilicus was normal. A midline fibrous raphe was noted running from the umbilicus to the epigastrium. Normal external male genitalia were seen.

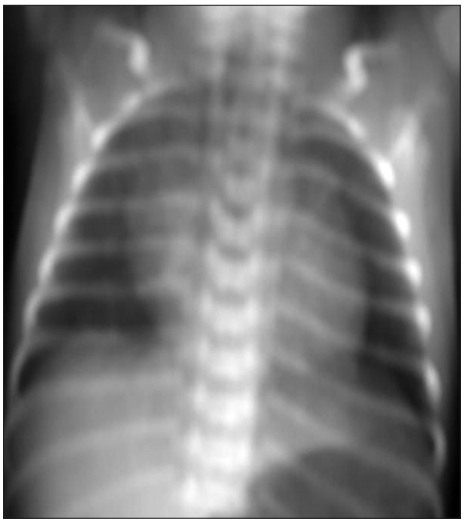

Figure 3: Chest X-ray showing centrally placed globular heart

A chest X-ray revealed a centrally placed heart, with normal lung fields. Echocardiography could not be done due to technical and financial constraints. The child was offered admission, with a view to have an early surgical intervention, but the parents refused this on financial grounds. The patient was lost to follow up after two clinic visits.

DISCUSSION

Cantrell's pentalogy (CP) is a rare congenital anomaly in which defects of the anterior abdominal wall, sternum, and diaphragm are associated with cardiac anomalies.[1-4]

Full pentalogy occurs rarely. Presence of the pentad is considered by some to be the full spectrum, with absence of some of the components seen as incomplete forms of the pentalogy.[5,6]

The prevalence of CP is estimated to be 5.5 in 1 million live births.[2] Its occurrence is probably sporadic.[6] CP is 2.7 times more common in boys, and African Americans may be more predisposed.[2] Two of our patients were females, while the third was a boy.

Pathogenesis of this syndrome has not been elucidated. The error in the embryology appears to occur in a segment of the lateral mesoderm. The cardiac abnormalities are due to a faulty development of the epimyocardium and the sternal and abdominal wall defects represent a faulty ventromedial migration of the paired mesodermal primordial structures. The diaphragmatic and the pericardial defects are secondary to a developmental failure of the transverse septum. These abnormalities seem to occur around day 14 to 18 of embryonic life.[2,7] Through the resulting sternal and abdominal wall defects, the organs may eviscerate.

Many factors may play a role in the pathogenesis of CP and ventral midline defects. Judging from the phenotypic variability of these defects, factors such as genetic aberrations and external mechanical forces may be implicated.[5]

Some workers believe that the pathogenesis of this syndrome can be linked to mechanical teratogenesis (band disruptions) following rupture of the chorion or yolk sac.[8] The mechanical teratogenesis concept seeks to explain the numerous malformations[5,6,9] that could be seen in these infants, exclusive of the torso.[8]

Toyama,[10] after a review of 61 cases of CP, suggested the following classifications: Class 1, definite diagnosis, with all five defects present; Class 2, probable diagnosis, with four defects, including intracardiac and ventral abdominal wall abnormalities present; and Class three, incomplete expression, with various combinations of defects, including a sternal abnormality present. Our three cases may be regarded as incomplete expression of the syndrome, as we were not able to demonstrate an intracardaic defect or a deficiency of the anterior diaphragm in any of them, due to constraints in performing echocardiography.

Successful corrective or palliative cardiothoracic surgery has been reported in these patients.[11] A multidisciplinary team approach should be employed in the management of these patients. The child should be reviewed by a pediatric cardiologist. Corrective cardiac surgery would be needed in patients with cardiac anomalies. Patients should be reviewed by a pediatric anesthetist before surgery, due to the peculiarities of anesthesia that may arise from the component anomalies.[7] We intended a repair of the sternal cleft in these patients, to provide a bony protection for the mediastinal structures. The technical and financial constraints we encountered, however, precluded this line of management.

The constraints we experienced in the management of these patients are rather sadly recurrent in pediatric surgical practice in this region, where a majority of the population survive on less than 1 US dollar daily,[12] and only about 2 – 3% of government expenditure is allocated to health.[13] Due to a non-encompassing health insurance, out-of-pocket private expenditure on healthcare is the norm. Therefore, healthcare is in direct competition with the basic needs of food, shelter, and clothing. This will explain the willingness of our patients' relatives to seek for discharge against medical advice on financial grounds. Policy makers

do not regard surgery as a component of basic healthcare and often dismiss pediatric surgery as being capital intensive and catering to an insignificant minority. This inclination has accounted for the paucity of equipments needed for the optimal care of our patients.

Cantrell's pentalogy is rare. Surgical repair of the sternal cleft should be done early to protect the mediastinal structures and eliminate a cosmetic deformity.[8] We recommend a multidisciplinary approach for its management.

REFERENCES

1. Cantrell JR, Haller JA, Ravitch MM. A syndrome of congenital defect involving the abdominal wall, sternum, diaphragm and heart. Surg Gynecol Obstet 1958;107:602-14.
2. Bittmann S, Ulus H, Springer A. Combined pentalogy of cantrell with tetralogy of fallot, gallbladder agenesis, and polysplenia: A case report. J Pediatr Surg 2004;39:107-9.
3. Di Bernardo S, Sekarski N, Meijboom E. Left ventricular diverticulum in a neonate with Cantrell syndrome. Heart 2004;90:1320.
4. Begum H, Nayek K. Cantrell's pentalogy. Indian Pediatr 2002;39:501.
5. Correa-Rivas MS, Matos-Llovet I, García-Fragoso L. Pentalogy of Cantrell: A case report with pathologic findings. Pediatr Dev Pathol 2004;7:649-52.
6. Polat I, Gül A, Aslan H, Cebeci A, Ozseker B, Caglar B, *et al.* Prenatal diagnosis of pentalogy of Cantrell in three cases, two with craniorachischisis. J Clin Ultrasound 2005;33:308-11.
7. Laloyaux P, Veyckemans F, Van Dyck M. Anaesthetic management of a prematurely born infant with Cantrell's pentalogy. Pediatr Anaesth 1998;8:163-6.
8. Groner JI. Ectopia cordis and sternal defects. Operative pediatric surgery. In: Ziegler MM, Azizkhan RG, Weber TR, editors. New York: McGraw - Hill Inc; 2003. p. 279-93.
9. Van Esch H, Mariën P, De Smedt M, Fryns JP. A boy with an unusual association of ventral midline anomalies including a trunk-like umbilicus. Clin Dysmorphol 2004;13:261-3.
10. Toyama WM. Combined congenital defects of the abdominal wall, sternum, diaphragm, pericardium and heart: A case report and review of the syndrome. Pediatrics 1972;50:778-92.
11. Acastello E, Majluf R, Garrido P, Barbosa LM, Peredo A. Sternal cleft: A surgical opportunity. J Pediatr Surg 2003;38:178-83.
12. Nwomeh BC, Mshelbwala PM. Paediatric surgical specialty: How relevant to African. Afr J Paediatr Surg 2004;1:36-42.
13. Ameh EA. Challenges of neonatal surgery in sub – Saharan Africa. Afr J Paediatr Surg 2004;1:43-8.

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