CASE REPORT

Pentalogy of Cantrell; complete expression in a nine-month-old-boy

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ABSTRACT

Pentalogy of Cantrell is a rare congenital malformation whose exact cause is not completely understood; it is characterized by defects in the anterior abdominal and thoracic walls consisting of omphalocoele, diaphragmatic defect, ectopia cordis, intracardiac defects and sternal clefts. The complex has variable clinical expression with complete and incomplete expressions reported. We, therefore, report a case of complete manifestation of the pentad in a 9-month-old boy.

Key words: Cantrell, ectopia cordis, omphalocoele, pentalogy, sternal cleft

INTRODUCTION

Pentalogy of Cantrell is a rare multiple congenital malformation which occurs worldwide with a reported incidence of 5.5 per million live births.[1] The exact cause is not known but is mainly thought to be sporadic, though its being associated with some chromosomal disorders like trisomy 18[2,3] and deletion on locus at Xq25-26 has been described in some cases.[4] It was first described by Cantrell *et al.,* in 1958[5] with the pentad consisting of a giant omphalocoele and a supra-umbilical anterior abdominal wall midline defect, sternal cleft, ectopia cordis, intracardiac congenital malformations like ventricular septal defect, atrial septal defect and tetralogy of Fallot, ventral midline diaphragmatic defect with defect of the diaphragmatic pericardim.[5] Other associated defects include cranial and facial anomalies, clubfeet, malrotation of the colon, hydrocephalus and anencephaly.[6,7] Pentalogy of Cantrell often have a poor outcome which is dependent on the severity of the malformations.[8] Though some cases of pentalogy of Cantrell had been reported in Nigeria, its true prevalence is not known and none of those reported had been proven to completely fulfill the five main components of the pentad.[9-11] We, therefore, report a case of pentalogy of Cantrell in a 9-month-old boy who completely fulfilled the diagnostic criteria.

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A 9-month-old boy was first seen at the age of 5 months being delivered with multiple congenital malformations involving the anterior abdominal and chest walls. The child was delivered at term gestation and at home; the pregnancy was not adversely eventful and she had no antenatal care. However, the child has defect on the anterior abdominal wall with a huge swelling extending from the lower anterior abdomen to the lower anterior chest wall, which was pulsatile and covered by a thin membrane which was initially reddish at birth but later became thickened and darker following daily cleaning and dressing. There was no history of vomiting or constipation, child sucked directly from the breast, though interrupted occasionally to catch his breath, there was no history of difficulty with breathing and no darkening of the lips or mucous membrane. The mother was a 25-year-old stay at home mother with four other children in a non-consanguineous marriage; she neither smoked cigarette nor drank alcohol and had no adverse medical record. No similar problem was found in the family. On examination, there was a big pendulous and pulsatile mass extending from slightly above the symphysis pubis to the epigastrum measuring 12 × 10 cm [Figure 1] with an epithelised membranous covering; it had visible peristaltic waves, it was pundunculated, with cardiac pulsation felt in the epigastric region [Figure 2]. Bowel and heart sounds were heard over the mass below the level of the diaphragm. There was a lower sternal cleft with absence of the xiphoid process. He was not tachypneic or tachycardic and had first and second heart sounds with a systolic murmur. Bowel sounds were audible and normal. His chest X-ray showed a midline heart with inferior displacement [Figure 3], abdominal ultrasound showed the swelling to contain loops of bowl and echocardiography

revealed a 6 mm secundum atrial septal defect. A diagnosis of pentalogy of Cantrell was made. The child is currently being followed up awaiting staged repairs of the defects.

DISCUSSION

Pentalogy of Cantrell is commoner in boys and less than 60 cases have been documented so far worldwide as at 2007.[8] The exact mechanism is not completely understood, however, the proposed pathogenesis involves a defect in embryogenesis between 14 and 18 days after conception due to failure of the in folding of the lateral mesoderm. The severity of this disorder is quite variable from one individual to another. While most affected children die shortly after birth due to the severity of their defect (especially the associated intra-cardiac defect and risk of infection in open defects), it has been reported in a 32-year-old-man.[12] Our patient was 9-month-old and remained active despite the associated atrial septal defect and has better prospect with adequately staged surgical intervention. Our case had all the major features of the pentad, though with a relatively favourable lesion because the membrane of the giant omphalocoele was completely epithelised [Figure 4] and the peduncle covered with skin, which helped in preventing infection; furthermore, the atrial septal defect was of moderate size and the child had remained stable, though he stands the risk of trauma to the barely covered heart, but with proper counselling the child has survived up till this age.

Pentalogy of Cantrell can be diagnosed prenatally but that has not been the case in those reported from Nigeria so far because lack of accessibility to a proper antenatal healthcare and for the fact that most of those diagnostic tools are not readily accessible to these low income parents.

CONCLUSION

Patients with complete expression of pentalogy of Cantrell are rare and few may survive to older age depending on the severity of the associated lesions, more especially the intracardiac defect. Furthermore, we advised that patients with giant omphalocoele should be thoroughly evaluated,

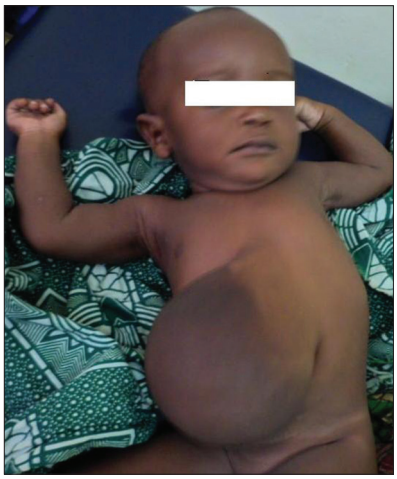

Figure 1: Giant omphalocele

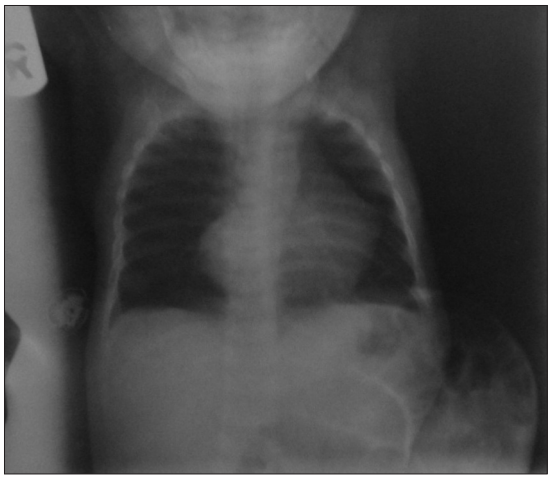

Figure 3: X-ray showing loops of bowl in the swelling with displacement of cardiac shadow

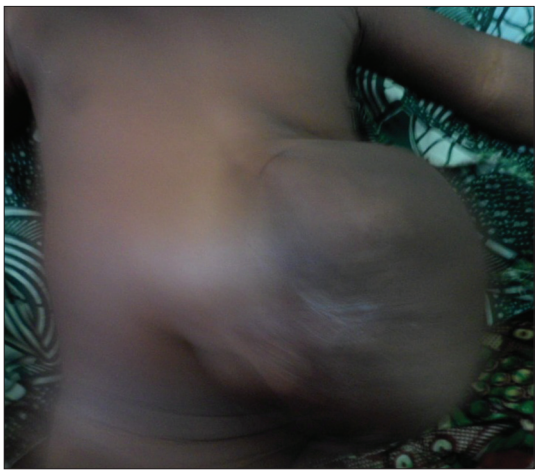

Figure 2: A bulge in the region of the epigastrium and lower sternum

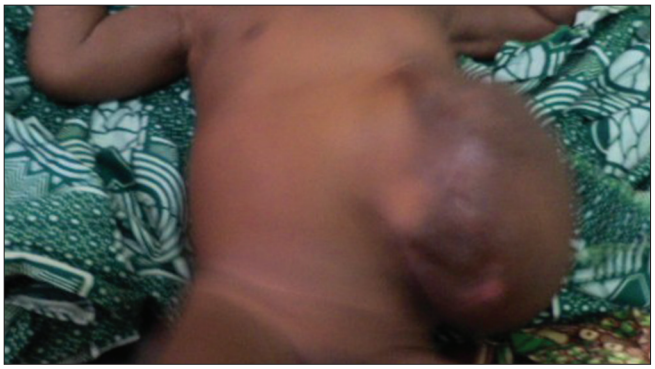

Figure 4: The mass covered by thickened, hyperpigmented keratinised membrane

because they may have an incomplete expression of the syndrome.

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